UNIVERSITY OF SOUTHAMPTON

General Practitioners, Generalism And The New Genetics

Qualitative and quantitative studies of general practitioners' responses to the implications of genetic advances in breast cancer for their roles

Satinder Kumar MB ChB MRCGP

A thesis submitted for the degree of Doctor of Philosophy

Department of Primary Medical Care
Aldermoor Health Centre
Southampton SO16 5ST

For my mother Kamla Rani
b.1936—d.1968

UNIVERSITY OF SOUTHAMPTON

ABSTRACT

FACULTY OF MEDICINE

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GENERAL PRACTITIONERS, GENERALISM AND ADVANCES IN CANCER GENETICS

By Satinder Kumar

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Policy-makers, clinical geneticists and the Royal College of General Practitioners recognise general practitioners will identify patients with genetic susceptibility to common cancers. They recommend most patients' enquires about genetic susceptibility to common cancers, such as breast cancer, should be managed by general practitioners (GPs). Since the ratio of consultant geneticists to general practitioners is roughly 500 to one general practitioners will necessarily have a role. This research explores GPs' ideas of their role and their responses to roles identified for them by policy-makers and experts. I used qualitative and quantitative methods. Grounded theory guided open-ended interviews with a purposive sample of GPs. Interviews explored GPs' ideas, beliefs and experiences of genetic medicine in general, and more specifically, in relation to breast cancer. As data collection and analysis proceeded a theoretical sample (selection guided by the emerging analysis) was generated to explore categories further, and to test the integrity and credibility of the emerging analysis. Two core themes were identified: 1) *genetics in the generalist context*, which included appropriate generalist intervention, the ethical dilemmas implicit in the therapeutic gap and the familial-hereditary distinction in primary care, and 2) *the implications for the generalist identity*, including the potential marginalisation of generalism. The category familial-hereditary distinction was used to construct a hypothesis and a questionnaire which was applied to a random sample of 200 GPs in the Wessex area. A descriptive statistical analysis and factor analysis of the questionnaire data supported the findings of the qualitative study. Both studies revealed inconsistencies between policy-makers' and GPs' definitions of general practitioners' role in implementing genetic advances. General practitioners' emphasised the need to build on current practice, whereas policy-makers focused on transforming practice to include new and specialised roles.

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In my reflections upon the state of my case since I came on shore on this island, I was comparing the happy posture of my affairs in the first few years of my habitation here, compared to the life of anxiety, fear, and care which I had lived ever since I had seen the print of a foot in the sand.....but I had never known it, and was incapable of any apprehensions about it; my satisfaction was perfect, though my danger was the same; and I was as happy in not knowing my danger, as if I had never really been exposed to it.....how infinitely good that providence is, which has provided in its government of mankind such narrow bands to his sight and knowledge of things; and though he walks in the midst of so many thousand dangers, the sight of which, if discovered to him, would distract his mind and sink his spirits, he is kept serene and calm, having the events of things hid from his eyes, and knowing nothing of the dangers which surround him.

Daniel Defoe

Robinson Crusoe (1719)

Chapter One

Introduction and overall aims

1.1 Introduction

The aims of this chapter are to introduce myself and the research described in this thesis. My research focuses on two main areas: the implications of genetic advances for general practitioners (GPs) in the context of identifying people with a genetic susceptibility to common cancers (e.g. breast cancer), and the types of knowledge required by general practice as a discipline—a dimension which emerged during the research process. I begin by explaining why, after completing my hospital and general practice training, I decided to enter academic general practice and become a PhD student. I then clarify why I chose *the new genetics* as primary care had been identified as a key site for the delivery of genetic services.

1.2 Entering academic general practice

After graduating from the University of Liverpool in June 1987 I spent the next five years training in hospital specialities: general medicine, general surgery, obstetrics and gynaecology, paediatrics, casualty, rheumatology, haematology and dermatology, as preparation for entering general practice training. The following year I worked as a locum doctor whilst waiting for the Central Manchester General Practitioner Training Scheme to advertise for their next intake of GP trainees. I

chose this scheme because of its reputation for being well organised, informative and supportive to trainees in transition from hospital practice to general practice. The scheme differed from many others because of its close links to the university of Manchester's academic department of general practice--the Robert Derbyshire Practice. Many of the tutors who lectured on the scheme were "academic" GPs undertaking research alongside their clinical practice.

1.3 General practice training—"a *revolutionary experience*"

Morrell (1993) described how doctors undergo "*a revolutionary experience*" when they move from the institutionalised environment of hospital practice--where doctors work in teams and clinical decision-making is shared-- to training and working in general practice which is a more solitary experience. In my own experience, a more significant difference between the two systems lay in their thinking and practice of medicine. The scheme I trained on encouraged trainees to adopt a critical approach, asking them to re-examine the professional and 'cultural values' they acquired during hospital training, and the problems of mapping practices learned in hospital-based training directly on to managing patients in general practice. Similarly, in the context of researching phenomena that constitute general practice, we examined how understanding context in general practice was key to explaining and understanding GPs' (and patients') behaviour and clinical decision-making. For example, research conducted by Bradley (1992), in Manchester, looked at GPs' prescribing decisions. He highlighted how decisions, which when assessed in terms of rational prescribing criteria alone appeared

irrational, seemed more reasonable once the clinical context of decision-making was understood. Bradley described how GPs experienced discomfort when they prescribed antibiotics for sore throats, because they knew they were acting contrary to the findings of research evidence. However, GPs explained their prescribing addressed patients' anxieties about their condition—a legitimate part of *holistic practice.* In this light, GPs' prescribing appeared not irrational but responsive to patients' emotional and psychological contexts.

During my year as a trainee, I was introduced to the works of Illich (Limits to Medicine-Medical Nemesis 1976), Neighbour (The Inner Consultation 1987) on the general practice consultation, and Armstrong (1984) on the doctor-patient relationship. That trainees were asked to engage with such works, suggested to me, general practice was a discipline ready to acknowledge, and keen to explore professional assumptions and processes upon which its clinical practice rested. As a consequence, I thought of general practice as open to new ideas, and to research methods beyond the experimental model. It was receptive to understanding, practising, and researching medicine differently—for me, this was revolutionary.

By the end of my year as a GP trainee I understood why general practice was considered to be a unique clinical discipline: its generalist focus, the gate-keeping role, the wide range of conditions managed in diverse contexts, its relative ease of access, and its provision of continuing longitudinal care (Starfield 1994). Added to these roles were pressures for practitioners' decision-making to be holistic, evidenced-based and economic (Heath 1995) --pressures which were at times in

conflict, giving rise to tensions between practitioners, patients and policy-makers. However, such tensions also acted as a force directing research into new developments in policy, clinical practices and technologies that claimed to offer improved clinical care.

Excited and stimulated by a year in general practice in which I had seen GPs operate as practitioners, teachers and researchers I decided to look for a post that combined research and clinical practice. In October 1994, I obtained a National Health Service (NHS) post as a half-time general practice principal in the Primary Medical Care Group at Southampton University. With this post came an opportunity to develop a research project with guidance from senior academic GPs and a medical-anthropologist based in the department. The project would form the other half of my post but needed funding, which came in the form of a South West NHS Research Studentship that funded for three years working half-time.

1.4 Selecting a topic for research

I was offered three options from which to develop a research project; 1) the management of otitis media in general practice, 2) the use of flexible sigmoidoscopy in general practice, and 3) exploring the impact of the new genetics on general practitioners. All three were relevant to primary care and could have made interesting, fundable and feasible research projects. The first two options related to specific clinical contexts. Otitis media is a common inflammatory condition of the middle ear which occurs most often as a response to infection.

This condition affects children more than adults and is a common reason for attendance in general practice. Its diagnosis, management and sequelae all have a significant impact on patients, parents and health professionals. A potential advantage to developing this option lay in the fact it was already an established area of research in the department, and so there was expertise on hand. The second option, the use of flexible sigmoidoscopy in general practice, focused on the feasibility of general practitioners using sigmoidoscopy as a screening and diagnostic procedure in patients over 45 years who presented with specific lower gastrointestinal symptoms e.g. rectal bleeding, or persistent mucus discharge. This would be a new area for research in the Primary Care Group and would serve to create links with the academic department of surgery at Southampton University. The third option was the impact of the new genetics on general practitioners--a far broader topic in comparison to the other two. By 1995, experts, policy makers and clinical geneticists were beginning to identify general practice as an important site for the delivery of anticipated developments in screening, diagnostic and therapeutics (Harris 1992, Austoker 1994, Genetics Advisory Group First Report 1995a, Calman 1995). Unlike the other two options, it was relatively under-researched in the context of British general practice.

Clearly, options one and two were important clinical areas for research that focused on GPs' management of common conditions in general practice. Such research, using trial methodology, had the potential to produce generalisable evidence that could influence: professional clinical behaviour, the development and use of interventions in general practice, and have implications for the organisation and

delivery of clinical care. For these reasons, funding-bodies may have looked upon research projects into otitis media and flexible sigmoidoscopy more favourably. In contrast, research into the impact of as yet undeveloped genetic technologies on general practitioners could have appeared too abstract and so potentially less appealing to funders. However, genetic advances generated a high level of public interest and debate at national and disciplinary levels, and in 1994 the new genetics occupied centre-stage in the public media and in academic journals from a broad range of disciplines. Moreover, debates published by research councils, policy-research institutions, clinical and non-clinical academic disciplines, and by parliamentary committees, agreed on the need for further research on the impact of genetic advances on society, from different contexts—and clinical practice and service delivery were flagged as priority areas (King's Fund 1994, Genetics Advisory Group 1995b, Calman 1995). The House of Commons Select Committee on Science and Technology (1996) also identified *professional responsibility of individual general practitioners to ensure they kept up to date with new procedures and developments in the field of genetics*. This statement echoed concerns about meeting patients' demands for advice on genetic susceptibility to common diseases. It was assumed that patients once aware of genetic advances would automatically seek services from the NHS.

Additional to the merits of each research topic were other more personal factors I needed to consider in choosing a topic. These related to the university's expectations that I would register for doctorate. I too was keen to use any research that was funded to form the core of a higher degree as I was keen to pursue a career

in academic general practice. Thus I took into account my own intellectual interests in the topic, as well as the degree of ownership I would have of the project. These were legitimate concerns as my studies would be part-time (six years for a PhD) and my funding was only for three years. So the topic I chose would have to sustain my enthusiasm and interest for up to six years.

Having spoken with all potential supervisors, and considered the factors outlined above I decided to develop option three, the impact of genetic advances on general practitioners. This was a novel area for research in the context of general practice, it held considerable interest for me and generated enormous interest in the public and academic arenas. Additionally, researching the new genetics offered an opportunity for me to learn about theories and methods from the social sciences which I had first encountered as a GP trainee. Given the relatively undefined nature of the topic in relation to general practice, it was clear that any research in to this area would involve using qualitative research methods (see chapters three and four).

1.5 Primary Care--a site for the provision of new genetic services

In 1995, advances in genetic science were clarifying the genetic basis of common conditions including common cancers e.g. breast cancer. Its was assumed that therapeutic, diagnostic and screening technologies would be rapidly developed using this new knowledge, and the expected patient demand for these technologies would require quick implementation in the health service (NHS Research and Development Report on the New Genetics 1994). This in turn raised questions

about who should provide genetic services in the future, and at what level?

The conventional model of genetic service delivery had developed around the diagnosis and management of relatively rare single gene disorders, which in general, were managed by regional clinical and laboratory genetic services (Harris 1992). This model would not necessarily be appropriate for managing common late onset disorders of low genetic penetrance. Further, it was anticipated that genetic advances would produce new diagnostic and therapeutic tools, e.g. genetic tests and drugs which would be relevant to the health of many individuals. Thus it was envisaged that future genetic services were more likely to be offered outside the regional centres e.g. primary care. This was made evident in number of policy reports. As early as 1989 a report from the Royal College of Physicians stated:

"GPs and others in primary care are in a particularly favourable position to recognise genetic problems and to utilise new developments in genetics. They, like, consultant clinical geneticists, are commonly concerned with the family as a unit, while their existing involvement with community based preventative measures, such as immunisations and cervical cancer screening, makes the primary care setting a logical one for genetic screening programmes."

In 1991 the House of Commons Science and Technology Committee's stated:....

"*...we believe that the weight of evidence (from the clinical genetics society) is sufficient to indicate a need for improved genetics services in the UK at the primary and community health care level."*

Five years later, the Chief Medical Officer, in his comments to the House of Commons Science and Technology Committee on Human Genetics (1996), emphasised general practice should not be excluded from developments in genetic science and that GPs would need to become familiar with genetic advances. Thus policy-makers and specialist services were clear about the necessity for primary care to be involved delivering future genetic services.

1.6 The potential role for general practitioners

In their second report to the NHS Central Research and Development Committee, the Genetics Advisory Group (1995*b*) recommended the role of primary care could include the following responsibilities:

- To assess the subject's preconceived ideas about the aetiology of disease
- Discuss perceptions of risk
- Construct a family pedigree
- Assess the risk of developing disease from the pedigree
- Help guide families to appropriate surveillance programmes
- Identify families eligible for genetic testing and refer to a specialist testing centre
- Identify and refer individuals who could benefit from psychological counselling

Austoker also defined a similar set of roles for GPs' in relation to genetic testing for common cancers (1994). Neither the Department of Health, nor Austoker

considered it necessary to explore how GPs' saw their role—GPs' opinions of what these should be remained unexplored. However, there was broad agreement between clinical geneticists, policy-makers and general practitioners (Quereshi and Raeburn 1993, Harris and Harris 1995) that even if primary care teams and general practitioners were willing to provide genetic services they were unlikely to possess the necessary information and skills in the context of common conditions. **What remained unanswered were questions about GPs' perceptions of the roles being defined for them, about how these roles related to current practice, and how they defined a role for themselves. These questions formed the substantive core of my research and were examined in the context of breast cancer.** As the work progressed additional questions emerged which centred on the types of knowledge required by general practice these questions are explored in chapter three.

1.7 Thesis outline

Following this introduction, chapter two reviews selected literature on the *new genetics,* from bio-medicine and social sciences to illuminate issues pertinent to GPs identification of people with genetic susceptibility to breast cancer—my chosen example of a common cancer. Chapter three, Shaping Knowledge, is a reflective chapter that focuses on the philosophical and theoretical underpinnings of qualitative and quantitative studies, and considers knowledge as evidence in general practice. Chapters four presents the grounded theory method used in the interview study of GPs, and chapter five details the qualitative analysis of the interview data.

Two themes are presented in this chapter: *genetics in the generalist context*, and *the implications of the new genetics for the generalist identity*. Chapter six focuses on the GP survey, its development, application and analysis of the data collected. The final chapter, (ch. Seven) provides space to reflect on the results on the studies, and allows a comparison of the findings with relevant literature published after the studies in this work had been completed. In this final chapter I develop a more theoretical discussion of the results informed by sociological and anthropological perspectives. The thesis ends by presenting the conclusions from this work, and by looking to future research opportunities.

Chapter Two

Literature review

2.1 Introduction

This chapter reviews literature on genetic advances in relation to breast cancer keeping in mind the roles identified for GPs by policy-makers, as detailed in chapter one. The introduction is followed by the aims and objectives of the literature review, which are addressed in three subsequent sections. The first section examines the idea of the *new genetics* from the geneticist's perspective, using the example of the polymerase chain reaction (PCR) to demonstrate the advantages brought to genetic inquiry by new research technologies. This section continues by examining a specific output of research in to the human genome: the identification of BRCA1, which is discussed in terms of its epidemiology, the risks it confers to carriers and their relatives, and the possibility of identifying carriers of BRCA1. The second section focuses on the implications this knowledge has for clinical practice, and specifically for general practitioners. The third section summarises the important issues identified by literature from the social sciences relevant to the general practitioner's role in identifying and advising patients about their genetic susceptibility to common conditions such as breast cancer.

In 1995, when I began this work, there were no published studies from British general practice examining general practitioners' perceptions of identifying people with a genetic susceptibility to common cancers. However, there was powerful rhetoric which called for GPs to receive more education and training in genetic advances to prepare them for the roles identified for them (Royal College of

Physicians 1991, Calman 1995, Austoker 1994, Harris and Harris 1995)—see box 2.1

Box 2.1. The roles identified for GPs by Austoker in relation to genetic testing for common cancers

- An awareness of the genetic dimension to common diseases such as cancers
- The dimensions of family history that are needed to assess genetic risk e.g. age of onset of the condition in the relative, the degree of relatedness etc.
- The scope of genetic testing for common diseases e.g. the benefits, limitations and risks
- Issues surrounding pre and post test counselling and continuing support for those at high risk
- Issues around recording sensitive information in notes/ ownership of genetic information
- Implications for insurance, employment and the family
- Where to refer and from whom to seek further expert advice
- How to counsel those who do not require referral

In 1995, published research focused on general practitioners' roles in screening for single gene disorders e.g. cystic fibrosis (Harris et al 1993). Although any one single gene disorder will be rare in comparison to other conditions managed in general practices, they are understood as genetic conditions by GPs, with clear rationales for identifying carriers, and screening of pregnancies (Harris and Harris 1995), which is not the case for conditions such as breast cancer. However, since 1997, studies examining general practitioners' perceptions of their role in identifying and advising people with genetic susceptibility to common cancers have

been published. In the context of this work, these studies were published after the qualitative study and the construction of the GP questionnaire were completed. Thus, they are examined in chapter seven in relation to the findings of qualitative and quantitative studies

2.2 Aims and objectives of the literature review

The aims of the literature review were to identify and examine the evidence, concepts, and assumptions integral to discussions about the GPs' role in identifying and managing people with genetic susceptibility to breast cancer. It was unnecessary in the context of my research to review genetics advances in great detail but it was important to arrive at an understanding that clarified the complexities that practitioners may face in practice. This was achieved through the following objectives.

1) To understand the genetic basis of cancer at the somatic level
2) To understand familial risk and relative risk of breast cancer
3) To understand the inherited basis of breast cancer
4) To examine the clinical implications of identifying major genes for breast cancer
5) To identify policy statements in relation to genetic advances and general practice
6) To understand broader social dilemmas practitioners may face in identifying and managing people with genetic risk

7) To gauge GPs' attitudes towards identifying people with genetic susceptibility to cancer

2.3 Evidence for the inherited basis of cancer

Evidence for genetic predisposition to cancer has a long history which precedes a detailed understanding of the human genome. Early breakthroughs in understanding the genetics of cancer were made in the context of relatively rare cancers e.g. chronic myeloid leukaemia and the Philadelphia chromosome (Noel and Hungerford 1960), and the gene responsible for retinoblastoma (Cavenee 1983). These advances identified genes which acted as tumour suppressor genes and oncogenes, and so clarified how mutations in these genes led to uncontrolled cellular proliferation. More recently, research conducted as part of the human genome project identified a number of highly penetrant genes predisposing to colon, breast and ovarian cancer and melanoma (Yates 1996). These genes are thought to be responsible for much of the cancer diagnosed before the age of 45 but cause a much lower proportion of these cancers seen in older people.

I have used the genetic advances made in the context of breast cancer to illustrate why there are pressures to prepare clinicians for implementation of findings in practice. I chose breast cancer because BRCA 1 had been identified in 1995. Since then other genes, e.g. BRCA 2, P53, and the atalexia-telengectasia gene have been identified as contributing to hereditary breast cancer. I decided not to discuss all of these genes as the issues and dilemmas raised by one gene, are on the whole, the same for other genes. My work focuses on BRCA1 to illustrate the issues GPs may

face, but I do discuss other genes when the heterogeneity of breast cancer genes has implications for clinical practice e.g. problems for screening, and complicating the estimation of risk for other people in the family.

2.4 Interest in the *new genetics*

To provided an idea of the interest the *new genetics* has generated I have included figure 2.1 which shows an increase in the number of articles published by leading bio-medical journals on genetic advances.

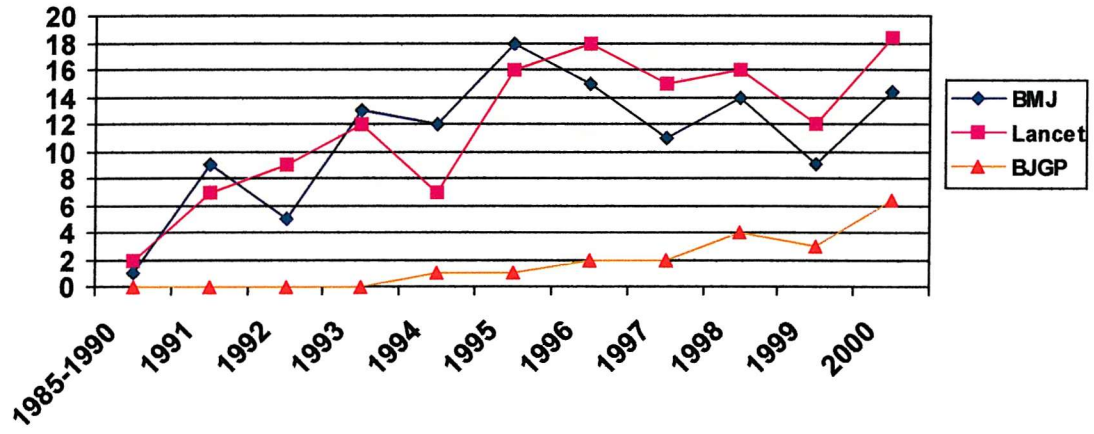


Figure 2.1 shows a 10 fold increase in the number of articles examining some aspect of genetic advances published by the British Medical Journal, with similar trends apparent in the Lancet and the British Journal of General Practice. Journals from the social and human sciences e.g. philosophy, ethics, law and sociology mirrored this trend shown by bio-medical journals.

2.5 The *New Genetics* –the geneticists' perspectives

According to Weatherall (1991), David Comings, editor of the American Journal of Human Genetics coined the term *new genetics* in his response to a paper that outlined a novel approach to using DNA analysis for mapping the human genome-- he commented:

"Since the degree of departure from our previous approaches and the potential of this procedure are so great, one will not be guilty of the hyperbole in calling it the New Genetics."

Here, Comings referred to the development of new techniques in molecular and cellular biology e.g. recombinant DNA procedures, gene cloning and DNA sequencing, which were made possible by the application of new tools such as the polymerase chain reaction (PCR). Important advantages conferred by the polymerase chain reaction were the simplicity and speed it introduced to DNA analysis. The simplicity PCR brought to DNA analysis was very important: prior to the development of PCR, researchers working on human genes required many years to master the techniques needed to work on DNA. With PCR, Markham (1993) states, a novice researcher could perform experiments and get meaningful results within a few days. As Tyrell (1997) explains, PCR allowed researchers to make multiple copies of any desired piece of nucleic acid. This amplifying effect allowed researchers to perform several examinations/experiments on any one sequence of genes simultaneously so accelerating the task of identifying and mapping genes.

The development of novel genetic research techniques: the *new genetics,* made sequencing the human genome feasible and achievable. The benefits of undertaking this were assumed to be substantial because they had the potential to improve understanding, management, and prevention of human diseases. Additionally, for governments who funded research in to the human genome the economic returns of developing this knowledge, e.g. in the context of treating and preventing disease, were predicted to be substantial (Garver and Garver 1994). Such arguments, were presented by many geneticists including Gilbert Smith, James Watson, Charles Cantor and Leroy Hood, when lobbying governments for large amounts of public funds to create an international collaboration of scientists who would map the human genome.

2.6 The Human Genome Project

The lobbying was successful and public funds were used to set-up the human genome project which was launched in 1990. Later this project became the Human Genome Organisation (HUGO), which co-ordinated the research of geneticists around the world. At its inception, the cost of mapping the human genome was estimated to be in excess of $ 3 billion (Jordan 1989). Garver and Garver (1994) put this sum in to perspective when he described the financial contribution made by the United States to the Human Genome project "*as the largest scientific project funded by the federal government since the Apollo Moon Project.*"

2.6.1 The aims of the Human Genome Project

The primary aim of the research co-ordinated by HUGO was to construct maps for each human chromosome at increasingly finer resolutions. The basic strategy for the project was:

1) To map all of the 50 000 to 100 000 human genes
2) To construct a physical map of the entire human genome
3) To determine the nucleotide sequence of all 24 chromosomes.

A map of the human genome would identify the location and sequence of genes, and the molecular markers and the spacing between them on each chromosome. The maps were to be constructed at different levels of resolution—the coarsest resolution was the genetic linkage map which would provide information on the location and sequence of genes and DNA markers. More detailed versions of the map would describe the chemical characteristics of the DNA molecule itself at each chromosomal location. Rapid progress was made in mapping the human genome and by 1992 complete physical maps of chromosomes Y and 21 and detailed RFLP (restriction fragment length polymorphism) maps of the X chromosome and all 22 autosomes were published (Yates 1996).

2.7 The genetic basis of cancer

Understanding genetic changes at the somatic cellular level were important in suggesting how common cancers, such as breast cancer, were inherited through the

germ-line. By the 1960s it was evident that cancer was essentially a genetic disease at the cellular level caused by abnormalities in genetic mechanisms which controlled cellular growth, differentiation and proliferation (Bodmer 1988). Individuals acquired alterations or mutations in their genes during their life-time through exposure to environmental carcinogens such as certain chemicals, radiation, infectious organisms, or factors in the diet (Bodmer 1991). Many carcinogens are mutagens: factors which damage DNA to produce alteration in genes. For example, the incidence of most solid tissue cancers, such as breast cancer, increases with advancing age, an observation which is consistent with the idea that cancer results from the successive accumulation of a series of genetic mutations (Knudson 1971). Statistical logarithmic analysis of cancer incidence with age of incidence estimates that at least 4-6 critical steps are involved in the development of malignant tumours, with each critical step taking between 10-15 years (Bodmer 1991). Each cancer can thus be regarded as an independent evolutionary process at the somatic cell level involving the usual sequences of mutation and selection but without the intervention of a sexual process. The accumulation of successive mutations that result in a cancer take place in step-wise fashion, with each mutation conferring a selective advantage and so leading to an expanded cell population. Another mutation can confer a further selective advantage in promoting more cellular expansion.

2.8 The inheritance of cancer: micro-evidence

If cancer is due to a series of genetic changes at the somatic level, then it follows that sometimes one or more of those changes may be inherited in the germ-line, and

so be present in every cell of the body at birth—this idea was first put forward by Knudson in 1971. An individual born with such a mutation would possess cells that had already progressed towards becoming carcinogenic. Knudson proposed that this "head start" could be the basis for a dominantly inherited cancer susceptibility. He suggested that a further genetic step which occurred somatically could be another mutation in the same gene, acquired during the life-time of the individual, which knocked out its function altogether. In other words familial inherited susceptibility to cancer acted as a dominant trait, but the effect at the somatic cell level was recessive. Thus, Knudson predicted cancer occurred due to genetic changes—tumour suppressor mutations--which were recessive at the somatic cell level suggesting functions which normally prevented the development of a cancer. Knudson's ideas have since been confirmed by the findings of research in to the inheritance of retinoblastoma and colon cancer due to inherited familial adenomatous polyposis coli, and more latterly breast cancer (Cavenee 1983).

2.9 The inheritance of cancer—macro-observation

Other evidence supporting the idea of cancer as an inherited condition came from three observational sources (Bishop 1999):

1) Existence of rare cancer syndromes which often demonstrate a Mendelian pattern of progression through generations in a family (e.g. retinoblastoma, palmar tylosis and oesophageal cancer).
2) Families containing a number of common cancers, where the number of such cases exceeds that predicted by population rates. Additionally these family were notable because they exhibited cancer at an early age.

3) At the population level an increased risk of cancer in relatives of cancer cases was noted; in many cases the relatives were at increased risk of the same cancer although relatives may also be at increased risk from other cancers.

The main difference between the three sources of observation is that the role of genetic susceptibility is clearest in (1) and least clear in (3). The importance of family history and clustering of cancer in suggesting a genetic susceptibility is explored below.

2.10 Familial relative risk and the inherited component of cancer

Breast cancer was recognised as having a familial component over 100 years ago (Broca 1866). Repeated observations that some families experience more than the expected number of cases of breast cancer suggested an inherited factor (Lovett 1976, Schildkraut and Thompson 1988). For example, epidemiological studies found that people with cancer gave a positive family history of cancer more frequently than controls and that the ratio of positive family histories was between two and four (Canon-Albright et al 1991 and Easton and Peto 1990). For women with a positive family history of breast cancer this would mean they were at an increased risk for developing the disease, but the magnitude of the risk identified differs in accordance with number and type of affected relatives, age at diagnosis of the index case and laterality. Additionally, different studies have identified varying estimates of risks which are a function of study design.

A measure of the contribution made by inherited factors is the *relative risk* of

cancer to first degree relatives when compared to the risk in the general population. For most cancers, the familial relative risks are of the order of 2-4 fold, but are higher for early onset cancers e.g. breast, ovarian and colon. For some cancers, such as testicular cancer and thyroid cancer, the relative risks are much higher at 9 fold--indicating very strongly the operation of inherited factors (Cannon-Albright, 1994). Moderate familial risks are sometimes taken to indicate there is little variation in cancer susceptibility but this argument may be fallacious as a relative risk of 2-3 fold could hide genetic effects of genes which have lower penetrance (see below).

Most of the familial risk observed in first-degree relatives of older cancer patients is probably not due to highly penetrant genes. A major unanswered question in cancer genetics is whether a substantial proportion of all cancers arise as a results of genes of lower penetrance.

2.11 Breast cancer

Breast cancer is the second most common cancer among women in the world; in developed countries it is the most common-- accounting for 500 000 deaths per annum (Hulka and Stark 2000). In the Western world the cumulative life time risk is 1 in 8 (American Cancer Society 1995). Rates of breast cancer are significantly higher in developed countries with the exception of Japan which has breast cancer rates half as high as those in North America and northern Europe (Pisani, Parkin and Frelay 1993). However, when women migrate from one country to another their breast cancer rate increases and assumes a pattern similar to the host country,

over 2-3 generations. This has been well documented in Asian-born migrants to the USA and to Asian-American women born in the USA (Stanford et al 1995). Factors associated with breast cancer include: family history, hormones (endogenous and exogenous), dietary (e.g. fat consumption), exercise level, alcohol, smoking, pesticides, biological factors (e.g. women in whom more than 75% of the breast has nodular densities on mammography are at increased risk), electromagnetic fields and ionising radiation. Of all of these factors, family history is considered as the most important risk factor (Houlston et al 1992). In the context of my work I will focus on *family history* for which breast cancer provides a good model for examining the complexities of family history for genetic susceptibility.

2.12 Familial and hereditary breast cancer

It is important to distinguish between the terms *familial* and *hereditary* breast cancer (Narod1994). Hereditary breast cancer is cancer which occurs in a woman who is believed to carry a mutation of a gene which predisposes to her to breast cancer (and ovarian cancer) e.g. BRAC1, and BRAC2. The presence of a constitutional mutation is inferred by the presence of extensive familial clustering (macro-observation), by direct DNA sequencing or by linkage analysis (micro-observation). If the cancer is due to a new mutation, or the woman's family is small, or the family history is incomplete (e.g. due to adoption of a family member etc.) then a woman with hereditary breast cancer may have no documented cases in her family, and may have no recourse to family history information.

The term familial cancer is often used by epidemiologists to signify a subgroup with

a family history of breast or ovarian cancer (e.g. one or more cases in first or second degree relatives). Familial breast cancer may be hereditary or simply be a chance occurrence, given the high frequency of breast cancer in Western populations. Thus there is likely to be a considerable degree of mis-classification if a single relative with breast cancer is used to define a predisposed sub-group.

2.13 Familial relative risk

Lynch and Lynch (1986) calculated that approximately 4% of cases of breast cancer were from families with multiple affected women and that the relatives of these cases were at much higher risk than quoted figures. The pattern of disease incidence in these families was consistent with an autosomal dominant inheritance (i.e. the risk for a woman with an affected mother or sister of carrying the gene reaching 50%) of a rare allele with a life time penetrance of 0.8 (first degree relatives of women in this subset would have an 80% chance of developing breast cancer during their life). Tulinius et al (1994) examined the implications of a family history of breast cancer for second and third degree relatives. They observed an increased risk for these distant relatives which was consistent with a genetic susceptibility. If breast cancer resulted from a gene at a single locus, and if the risks to first degree relatives was 2.26 then this would translate in to a risk to second degree relatives of 1.63.

The risk of breast cancer among relatives increases with early age of onset in the index case (a first degree relative). The relative risk for a sister was estimated to be 4.3 for breast cancer diagnosed in her relative at the age of 30, 2.7 for cases

diagnosed at age 40, and 1.7 for cases diagnosed at the age of 50 years (Claus, Risch and Thompson 1990). For index cases above the age of 55, Houlston et al (1992) estimated the increased risk to first degree relatives was in the order of 1.57.

These figures support the idea that some women develop breast cancer due to genetic susceptibility. Even though multiple cases of breast cancer will occur in families simply because it is common, early onset and relatives with bilateral breast cancer should alert clinicians to the possibility of a genetic predisposition—observations that could be used by general practitioners to guide the process of identification.

2.14 Understanding familial risk factors

Risk, in a epidemiological sense, describes a person's increased risk for experiencing an event when compared with the general population (Adams 1995). This is often difficult to translate in to the real risk an individual may face. For example, a 30 year old woman, in the UK, whose mother and sister have had breast cancer has a 43-fold greater chance of breast cancer compared with the general population. However, this does not simply mean that a woman has a life time risk which is 43 times that of the general population risk (1 in 12 in the UK) because the relative risk is specific only to a particular age. So, in the general population the relative risk of a woman developing breast cancer at the age of 30 years is 1 in 8000; 43 times this risk is 1 in 186. However, once this woman has reached 60 years of age and is unaffected by breast cancer then her risk reduces to very near that of the general population for that age. This is because any strong genetic

tendency should have expressed itself by 60 years of age (Eeles 1996).

A more useful and clearer concept of risk that could be used by general practitioners and patients is that of cumulative risks. For a example, a woman may want to know her risk for developing breast over her life-time, over a fixed period of time or until she is 60 years of age. Cumulative risks can be estimated from epidemiological studies in which the incidence of breast cancer in women with one or more affected relatives is compared with the incidence in women without affected relatives. The most comprehensive data in this field are from the Cancer and Steroid Hormone Study Group in the United States (Claus et al 1991). Graphs of the cumulative risk of breast cancer are readily available and could be a tool GPs use when explaining risk to patients.

2.15 The genetics of breast cancer

Once the pattern of distribution of breast cancer in families had been described it was clear that a dominant gene or genes model could account for at least 4% of breast cancer cases. In 1990, Dr Mary-Claire King et al, using linkage analysis, identified a locus on the long arm of chromosome 17 as having a substantial role in early-onset breast cancer (King 1992). Little evidence was observed for linkage in late onset breast cancers. An international consortium of geneticists, consisting of 13 groups working in 8 countries, used King et al's linkage-analysis to mark BRCA1, a large gene on chromosome 17, as the most likely candidate based on the mapping results of 214 families (Easton, Ford and Peto 1993). BRCA1 was finally identified in 1994 (Mikki, Swensen and Shattuck-Eidens 1994, and Futreal, Liu,

Shattuck-Eidens, 1994). A second locus, BRCA2 was mapped to chromosome arm 13q and appears to account for a proportion of early onset breast cancer roughly equal to that caused by BRCA1(Wooster et al 1995).

Since 1995, other genes conferring susceptibility to breast cancer have been recognised and include HRAS 1 an oncogene, A-T gene (gene for ataxia-telangectasia) and P53. These genes confer risks which would rarely produce large family clusters of cancer but could be responsible for a significant proportion of breast cancer in the population. In the context of general practitioners identifying women with a genetic susceptibility based on family history, it is likely most breast cancers due to these genes would be mis-classified as sporadic.

2.16 Gene frequency of BRCA1

Claus et al (1991) estimated the frequency of a dominant breast cancer susceptibility gene in the population to be 0.0033,or roughly 1 in 150 women is predisposed, a figure which represents the sum of the frequencies of all the dominant genes associated with breast cancer. However, it is now known that only 60% of hereditary breast cancers are linked to BRCA1. This realisation together with Easton, Ford and Peto's (1993) estimate of the frequency of alleles to be 0.0007 (based on 44 breast cancer deaths occurring among relatives of 1203 cases of ovarian cancer) puts the frequency of carriers of BRCA1 mutations in the population at closer to 1 in 500. However, a more recent analysis of British data estimated the frequency at 1 in 833 (Ford, Easton and Peto al 1995). From this it is clear that estimates for the prevalence of BRCA1 vary in Western populations--

British data (Ford, Easton and Peto 1995) suggested the prevalence was 1/833 and American data (Claus et al 1991) 1/150, why this is so is not yet clear but explanations may include differences in the ethnic compositions of populations or the effect of exposure to differing levels of mutagens. Current studies show that breast and ovarian cancers due to BRCA1 in the general population are low. Whittemore et al (1997) estimated that 4.2% of all breast cancers (and 5.3% of all ovarian cancers) diagnosed before the age of 70 years were due to BRCA1.

2.17 Penetrance of BRCA 1

The relative risk for breast cancer among BRCA1 carriers varies from 100 for cancer below the age of 30 years to 1.6 at 75 years. Based on a gene frequency of 0.0012 (1/833) for mutant BRCA1 alleles, this implies that breast cancers due to BRCA1 varies from 28% below the age of 30 years to less than 1% above the age of 70 years. It is estimated that BRCA1 accounts for approximately 45 % of families with several cases of breast cancer, (King 1992), and in up to 67% of such families the age onset was less than 45 years.

2.18 Germ-line BRCA1 mutations

More than 100 mutations have been identified in the BRCA 1 gene. However, only a small number of these mutations have been observed to occur repeatedly. Of particular interest was a specific type of gene mutation that appeared in more than 20 Jewish families with familial breast or ovarian cancers. Recent population surveys including one of Ashkenazi Jews, revealed that approximately 1 per cent

carry this mutation—apparently derived by descent from a common ancestor (Langston, Malone and Thompson 1996 and Fitzgerald, MacDonald and Krainer 1996). This is a very high frequency making breast (and ovarian cancer) attributable to 185delAG mutation a common and serious single-gene disease affecting a specific population group. The frequency of germ-line mutations in Jewish women is estimated to be in the order I in 107 for the 185del AG mutation. Using these figures, and the age-dependent penetrance curves for the observed risk of breast cancer in families with BRCA1, it was calculated that 7.5 % of non-Jewish women and 38 % of Jewish women with breast cancer under the age of 30 years would be expected to have germ line BRCA1 mutations (Struewing, Abeliovich and Peretz 1995, and Ford, Easton and Peto 1995). Thus, for this group of people there is the possibility of developing screening tests for the 185delAG mutation. Based on the results of which women may be offered the prophylactic option of surgical intervention (bilateral mastectomy and oophrectomy) before the disease develops. More controversially, the option to identify fetuses which carry this mutation and offering parents selective termination had been put forward (Lerner 2001)—thus treating this condition in a similar manner to Huntington's disease. However, there is evidence not all women who carry this mutation will develop cancer in their life time. This separates 185delAG mutation from gene mutations responsible for conditions such as Huntington's disease, where if the gene is present then the disease will always manifest. However this has not stopped physicians offering selective termination to women who carry 185delAG.

It is important to recognise that whilst genetic research has identified several genes, which display Mendelian models of inheritance much less is known about how the

function of these genes is modified by environmental factors or the presence/absence of other genes. For example, it is still not clear why between 15-20% of women who carry BRCA1 do not develop any of the cancers associated with this gene. Further, it is hypothesised that the majority of breast cancer cases may be due to interaction between genes with low penetrance and environmental factors. Thus, individual genetic variants would have little predictive power so the determinism attributed to genetic factors can not be taken for granted (Lewontin 2000).

2.19 Genetic heterogeneity and environmental factors

The heterogeneity of the BRCA1 mutations, underline the considerable challenge to developing a pre-symptomatic genetic test: the analysis of a specific gene, for identifying women at risk of breast cancer. Currently, in the context of the general population, no one test will reveal the presence of all mutations in BRCA 1 that confer susceptibility to cancer. Thus a gene test that is negative is only negative for the precise mutation it was developed to detect (Narod 1994). It is possible the woman may carry a different mutation, or even different genes conferring susceptibility. Additionally, her risk after a negative genetic test will be the minimum population risk. Thus GPs providing the post-test advice to women who test negative will need to make this clear, that a negative test does not mean risk free.

2.20 Reliability of family history as a predictor of genetic susceptibility

The evidence presented above, for germ-line mutations causing breast cancer, illustrates the potential of identifying women with susceptibility to breast cancer. However, family histories even for people who carry a mutation such as BRCA1 may be misleading. For example, studies by Langston, Malone and Thompson (1996) and FitzGerald, MacDonald and Krainer (1996) showed that many women with breast cancer had a first and second degree relative with breast cancer but relatively few *"had dramatic family histories that would have been considered highly suggestive of hereditary breast cancer."* Thus from these studies a striking family history can not be relied upon to identify all possible carriers of breast cancer.

BRCA1 mutations can be inherited through the male-line. Father's of women with BRCA1 have a moderately increased risk of prostate cancer but they often remain cancer free. Thus, a women who develops breast cancer due to BRCA1 inherited from her father may not have an affected first-degree relative, or affected second degree relatives unless there are paternal aunts who developed breast cancer. Moreover, some family members with BRCA 1may fall in to the 15-20% who never develop breast cancer. In these ways BRCA1 can remain silent in families, so when breast cancer occurs it may appear to be sporadic. At this point, the age of the women and bilateral disease may alert GPs to the possibility of genetic susceptibility and so potential risk to other family members.

2.21 Problems with recognising family histories as indicating genetic susceptibility

Central to the roles identified for GPs (see section 1.6) is identification of individuals whose family history suggests genetic susceptibility to cancer. As highlighted above, there are problems associated with using family history which may thwart recognition. Breast cancer is a common condition so any one family may have several cases by chance. Thus to achieve a greater level of discernment GPs would need to ask about the ages at which family members developed the cancer, if there was a history of bilateral disease, and the incidence of other cancers associated with BRCA 1 (e.g. ovarian, prostate, endometrial and colon cancer). Once this was established the GP would need to know how related the patient was to family members with cancer. Patient are known to find clarifying their relationships with more distant family members difficult and may require guidance (Michie, McDonald and Marteau 1996, Emery, Kumar and Smith 1997). Additionally, patients may themselves be un-informed of disease patterns beyond immediate members of their family, and even if they can identify and access other family members they may be reluctant to raise the issue of cancer. Once it is decided that a family may have a genetic predisposition, the second problem is to estimate the risk to individual family members.

BRCA1 and breast cancer predisposition are thought to be dominantly inherited, so that on average, only half the children of gene carriers will carry the gene. However, there are no reliable markers for susceptibility to breast cancer so that assigning very high or very low risks to specific family members will be difficult. If individuals and families are assigned to be high risk the third problem is deciding

what needs to be done about it, since, as mentioned previously, 15-20% of such carriers will remain cancer free. It could be other gene(s) provide a protective effect, or it may be the effect of lifestyle or environment. Clarification will be provided by epidemiological research once large numbers of BRCA1 carriers have been identified and studied prospectively (Evans, Cuzick and Howell 1996). Currently, it is not known whether factors known to influence non-sporadic cancers: age of menarche, contraceptive pill use and tamoxifen have the same importance for BRCA1 carriers. It could be that the genetic mutation over-rides the effects or that carriers are sensitive to one or more of these factors.

2.22 Family history collection and recording in general practice

So, how effective were GPs at recording family history and at identifying families who were related to one another? How knowledgeable were GPs about each family on their lists? How systematic was their recording of the physical, social and psychological factors? Did they collect family history to assess the genetic susceptibility to common conditions such as cancers of the breast, ovary and colon? Research addressing such questions was mainly conducted prior to the computerisation of general practice, and before the contractual changes of 1990s which coerced GPs to collect a limited family history of common chronic diseases such as asthma, diabetes, hypertension, ischaemic heart disease and stroke.

Much of the early literature from general practice on family history focused on developing systematic methods for recording family history that would facilitate the GP with social and emotional insights (Jameson 1968, Cormack 1975, Zander

1977, Cole 1978). For example Jameson (1968) designed a card index to record the names and relationships of people in a family as well as *diseases, symptoms and social history relevant to the family.* The aim of the card was to improve the continuity of care and allow the doctor to observe conditions or individuals which caused the family most problems. General practitioners were expected to understand how social context influenced patients' health seeking behaviour, life-styles as well as the diagnosis and management of symptoms. Further, because GPs provided continuing care to individuals and families they were in a position to observe patterns of disease and illness, health seeking-behaviour, socio-economic experiences, that occurred in a family over many generations. All this was identified as part of GPs provision of holistic clinical practice.

Cormack (1975) studied 187 GP hand-written notes and found that in no instance was family history recorded in a systematic way. Family history tended to be scattered throughout the day-to-day notes, which made it less useful because it was not easily accessible during consultations. Zander examined the extent of the GP's knowledge of family and social history and suggested that it may be less substantial than many would care to admit (Zander 1977). He showed that many GPs' personal knowledge of family history was lacking to the extent that there was uncertainty about the existence of 46% of close family members, although he did not clarify how he defined "close." More recent evidence from a questionnaire study (Summerton and Garrod 1997) suggested GPs continued to use family history as an insight in to the social and psychological context, and to aid decision-making about common conditions such as asthma, but whether this reflected exploration of shared environment or genetics was not made clear. However, what was evident from

Summerton and Garrod's work was GPs' knowledge of families, their relationships, and the use to which family history was put was erratic. A view echoed by an occasional paper, Genetics in Primary Care (Royal College of General Practitioners 1999).

Thus the evidence available on family history collection in general practice suggests GPs are not yet recording family history in the detail required to assess genetic risk of cancer such as breast cancer. Family history continues to be used as a source of social, psychological and life-style information about individuals and families. Its potential as a source of concrete genetic information for common conditions was not realised. In the United States education and training programmes have been introduced for family physicians focusing on the use of genograms, which are similar to family pedigrees used by geneticists (Waters et al 1994). This approach has also been advocated by in the UK by Emery and Rose (1999) and is presented as a way of integrating biological and psycho-social family history. However, what is not made clear is how using genograms will achieve such integration and the term itself is problematic because it refers only to the individual's/family's genetics.

Enquiring about and recording family histories of common conditions such as breast cancer will raise a number of social, psychological and ethical dilemmas for general practitioners. They are examined below by reviewing some of the critiques of genetic advances from the social sciences. However, before progressing to those arguments I have summarised below the assumed benefits of genetic advances to health.

2.23 Clinical implications of sequencing and mapping human genes

It is assumed that mapping the human genome will help identify genes contributing to, or responsible for single gene disorders, and perhaps more importantly common conditions. However, information about the genes involved in disease aetiology will have greater meaning once the mechanisms by which genes control biological processes, and interact with environmental factors are discovered. Thus, the identification and location of human genes is only the start of a process aimed at analysing gene function, and interaction of gene products. Once more comprehensive knowledge is available, its impact it is assumed, will be on the development of therapeutic and diagnostic materials (Bell 1997). The future benefits of genetic advances are:

- It will clarify the genetic components of common multifactorial diseases such as ischaemic heart disease, depression, diabetes and common cancers
- It will lead to a clearer understanding of the underlying patho-physiology and biochemistry involved in the aetiology of the disorder
- It may lead to the development of genetic tests to detect the carriers of genes conferring increased susceptibility to common diseases, providing the opportunity to prevent serious diseases or institute earlier, more effective treatment
- The development of new therapies targeted at the underlying molecular disorder and specifically designed to suit the genotype of the individual (Report of the Genetics Research Advisory Group 1995 *a*)

Bell has also asserted that within the next decade genetic testing will be used widely for predictive testing in healthy people and for the diagnosis and management of patients. Such predictions have been described as *a discourse of hope or great promise* (Durant, Hansen and Bauer 1996)—which include the most optimistic views about the potential power and benefit of genetic advances. According to MacIntyre (1997) the most optimistic accounts of genetic testing and screening are to found in official reports and not in the tabloid press—with the most optimistic account of benefits given by the NHS Central Research and Development Committee.

2.24 Social and psychological issues associated with clinical genetic advances

In response to this optimistic view of genetic advances other authors took a more cautious view of developments and identified some of the legal, moral, ethical, social and philosophical issues raised by genetic advances--highlighting some of the difficulties and detrimental effects of genetic screening and testing. This is *the discourse of concern* (Durant et al 1996) which can be summarised as

a) geneticization of society—in which differences between individuals are reduced to differences between their DNA (Lippman 1991)
b) underestimation of the importance of environmental factors
c) discrimination, stigmatisation (in insurance, employment and more generally)
d) changed attitudes to parenthood and commodification of babies
e) diversion of care, treatment and resources away from disabled people or people carrying genetic mutations

f) screening for conditions, like breast cancer, for which there is no effective treatment or surveillance

g) uncertainty whether life-style changes would occur as a result of screening, and whether they would be effective.

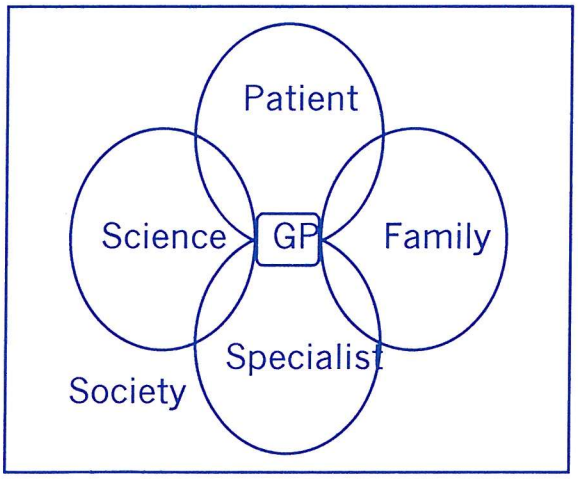
MacIntyre (1997) suggests these issues are not new and have been discussed previously in relation to screening for other conditions e.g. Huntington's disease. However, for GPs many who will rarely encounter conditions such as Huntington's disease these issues may be novel.

2.25 How are these discourses relevant for general practitioners?

General practice has been identified by health policy makers and clinical geneticists as occupying a pivotal position for the future successful application of genetic advances to society. As the point of first contact, general practice will be the site where the majority of enquiries about genetics occur (Harris and Harris 1995). Moreover, it is evident that in the United Kingdom, existing regional genetic centres, which serve populations between two and five million people, would be unable to deal with any increase in demand for genetic advice from the community. For example, the ratio of general practitioners to consultant geneticists has remained at 500:1 (RCGP 1999 and RCP 1999 personal communication). Policy makers and geneticists tend to emphasise these facts without acknowledging some of the more powerful and positive reasons for why GPs should be involved in delivering future genetic services.

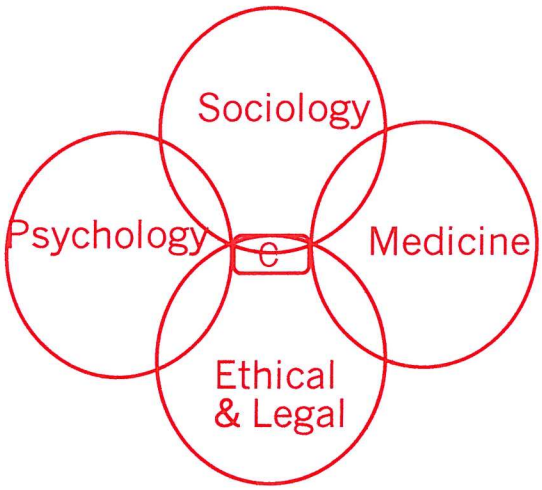
Within medicine general practice places greater emphasis on practitioners to make diagnoses in the context of the patient's physical, psychological and social conditions (Pendleton 1984). Post graduate general practice training schemes are organised to ensure that GPs possess a diverse range of consultation skills drawn from clinical medicine, and informed by sociology and psychology as well as, an understanding of ethical and legal matters. Furthermore the Royal College of General Practitioners advocates that GPs should also consider the broader implications of a consultation, balancing their role of patient advocacy with the needs and attitudes of society, the scientific community and specialist clinicians (Royal College of General Practitioners 1996). Figure 2.2 illustrates the boundaries of general practice and its central process, the consultation.

Figure 2.2 a) The unique position of general practitioner in delivering health care



GP = General practice

Figure 2.2 b) The unique position of the general practice consultation



C= Consultation

It is evident that general practitioners possess many of the skills important for managing the issues raised by the new genetics at the level of the individual and the family. For example, patients regularly present their GPs with newspaper cuttings or with information about specific scientific or medical discoveries. GPs manage such situations by acting as a mediator between science and the individual, facilitating the patients understanding of the particular scientific or medical advance presented. During consultations GPs sometimes explain the science behind an advance, particularise it to the patient's case and clarify realistic medical outcomes. It is this experience of mediation and of being bounded by individual, family, and society that may allow GPs to effectively mediate between discourses of promise and concern. GPs will need to balance promises and concerns when deciding to identify patients with genetic susceptibility, and when advising patients of the implications identification may have.

2.26 Summary

In this chapter I reviewed the genetic advances made in relation to breast cancer to illustrate how this new knowledge is creating pressures to re-organise future health services. This pressure is manifest in the roles being identified for GPs by policy makers and experts. The chapter summarised the type of genetic knowledge that GPs will need to fulfil the role of identifying and managing patients' genetic susceptibility. Specific issues relevant to the incorporation of scientific advances into clinical practice were identified and discussed: the probabilistic nature of *the new genetics*, the complexity of genetic knowledge, the details of family history required to assess genetic risk, and the limitations of family history as an indicator of genetic risk were identified and discussed. Finally, genetic advances were discussed as discourses of promise and concern, ideas which GPs may have to discuss when explaining the implications of being a carrier of genes associated with breast cancer. GPs ability to mediate between different types of knowledge (e.g. lay and professional knowledge, individual and family) was identified as a powerful reason for why GPs may be well positioned to provide advice about genetic advances to individuals and families.

In the next chapter I will review the theoretical and methodological issues raised by researching GPs' perceptions of the roles advocated for them and their own ideas of what their role should be.

Chapter Three

Shaping knowledge

3.1 Introduction

This chapter was started at the outset of the whole project and over its duration, the content has developed to reflect the methodological, theoretical and philosophical issues inherent in undertaking this type of research. The content, focus and structure of this chapter was shaped and re-shaped in response to new questions raised by different phases of research and writing-up. What these questions were and how I engaged with them are presented here. So, in one sense, this chapter illustrates how I constructed my understanding of qualitative and quantitative research, their relationship to each other, and how they operated in general practice research. Some of the issues I discuss relate to the construction and meaning of "knowledge" in medicine, and specifically in general practice. Such issues are not traditionally addressed in medical undergraduate or postgraduate curricula, nor are they well developed in general practice literature. In the context of executing and articulating this research, I became increasingly convinced of my need to understand the theoretical, philosophical and pragmatic differences between qualitative and quantitative methods and knowledge. Ultimately, it was securing this understanding and knowledge that allowed me to bridge the divergent assumptions underpinning qualitative and quantitative research and so understand how each method produces legitimate but complementary forms of knowledge and evidence. Writing this chapter was part of a process that equipped me to undertake qualitative and quantitative research, understand how to link qualitative and

quantitative studies, and develop a capacity to theorise the research findings. In this sense, this chapter provided a theoretical foundation for the thesis.

The specific areas addressed in the chapter are listed below:

a) the construction of knowledge in general practice

b) the philosophical position of this thesis

c) my rationale for using qualitative and quantitative methods

d) meaning of research paradigms

e) philosophical assumptions of qualitative and quantitative methods

f) defining qualitative research

g) the tensions in using qualitative methods in academic general practice

h) the researcher in qualitative research

I was aware that qualitative research methods and qualitative research were viewed with scepticism by many quantitative researchers within my own discipline. The reason most often cited to support this position was that qualitative research did not fulfil the criteria of "good science." As I intended to use qualitative and quantitative research methods, I wanted to make my own assessments. As a clinician, with a background in science I was virtually unaware of qualitative research and its methods but had a good understanding of the scientific method. During the year it took to convert the original research idea into a concrete and funded project (October 1994-0ctober 1995), I read books on the philosophy of knowledge, (Sorrell 1994, Losee 1993 and Trusted 1997) the philosophy of social research (Hughes 1996) and the scientific method (Gower 1997). Importantly, I was able to

discuss my reading with my supervisors one of whom was a medical anthropologist cognisant with the philosophical and theoretical foundations of qualitative research.

I began by familiarising myself with qualitative research published in journals such as *Qualitative Health Research* (e.g. Brock 1995) and *Social Science and Medicine* (e.g. Blaxter1983). I compared this work to qualitative research published in biomedical journals such as the *British Medical Journal* (e.g. Green 1993). There were significant differences between the papers in the *Qualitative Health Research Journal* and *Social Science and Medicine* to those in bio-medical journals e.g. the BMJ, JRCGP. Overall papers in biomedical journals were more likely to provide procedural clarity but exhibited limited analysis and theoretical abstraction of the data. Additionally, each paper devoted substantial space to convincing readers why the research was legitimate. In academic general practice there is a strong rhetoric promoting the use of qualitative research where appropriate, however in many instances the acceptance and deployment of qualitative methods was limited by the discipline's lack of familiarity with qualitative theory (Hoddinott and Pill 1997). This is evidenced in the relative, though not total, absence of theoretical exposition and clarification that characterises qualitative papers in bio-medical journals. However, constraints to theorisation also arise from disciplinary/intellectual and practical (editorial/journal style) factors. Restricting theorisation, these factors promote a formulaic approach to the presentation of qualitative research by those outside disciplines in which qualitative research is the primary focus. This lack of attention to theory, may lead to decisions about sampling, data collection and analyses that are driven by practical issues so jeopardising the credibility of the knowledge created (Harding and Gantley,1998).

3.1.1 Personal aims and objectives

In accordance with the conviction in qualitative research that the researcher is the research instrument, (Cassell 1977, Guba and Lincoln 1981) and should therefore disclose agendas at the outset, this chapter proceeds through an initial statement of my objectives and aims. With specific reference to my own research, this chapter serves the objectives of methodological clarity and credibility listed below:

1. to move with clarity between the different theoretical assumptions of the methods used
2. to prevent a formulaic approach to qualitative research (Harding and Gantley 1998)
3. to ensure my analysis of the qualitative data would be theoretically driven
4. to enable me to link the qualitative and quantitative data within the limits of boundaries set by their differing philosophical constraints
5. to underline the distinct nature of, and different limitations to qualitative and quantitative knowledge
6. to defend qualitative research methods to my peers in academic general practice concerned by issues surrounding their credibility
7. to clarify why I used both methods to research the implications of genetic advances
8. to contribute to the debates in general practice and bio-medical journals on the role and development of qualitative research methods in academic general practice

9. to facilitate the process of increasing my *theoretical sensitivity* (Glaser 1978) during data collection and analyses of the qualitative data (see chapter 4).
10. to aid *manufacturing distance* (see chapter 4) (McCracken 1988) from the familiar assumptions about the scientific method, and those shared with other general practitioners.

3.1.2 Needs of my peers

A further need to include this chapter was highlighted by my peers. Their unfamiliarity with qualitative knowledge and evidence became evident as the interview study (see chapter 5) was presented at local conferences (South West Association of University Departments of General Practice 1995), at national conferences (Association of University Departments of General Practice 1996 and the Royal College of General Practitioners Annual Research Day 1996) and international conferences (British Medical Association Annual Clinical Conference, San Francisco 1997, International Qualitative Health Research Conference, Vancouver 1998). Examples of the enquiries I recorded in my field notes were; "How can qualitative studies be useful, your sampling is too selective, you can't generalise your findings?" and " How can qualitative research and genetics go together! ...I thought it was all about molecular science and genes?" and " Why do you use all these '–isms? I might be able to understand qualitative research if it didn't use all that jargon." The questions suggest unfamiliarity with qualitative research, not only in terms of its language, but the theoretical assumptions that define its aims and scope for contributing to the knowledge base of general practice.

3.2 The construction of knowledge in general practice

The clinical discipline of general practice has existed in some form for several centuries, in contrast to the more recent academic discipline which was established in 1956 with the creation of the first university department of general practice in Edinburgh (Freeman 1992). The 1960s witnessed an expansion of academic departments in Europe and North America, and research in these departments began to establish a body of knowledge with a distinctive general practice perspective. Current British academic general practice aims to rest on three interrelated activities: practice, research and teaching. The need to improve clinical practice and clinical outcomes links all of three activities. However, despite the proliferation of academic departments, general practice as an academic discipline has recently been described as "*a*theoretical" (Howie 1998). Whilst this description is itself ultimately problematic -- because no research is without its theoretical backdrop, whether or not this is made explicit -- the characterisation is understandable, given that general practice and its research are applied areas with practical goals. Nonetheless, current general practice research could be better characterised not as *a*theoetical but *poly*theoretical, in so far as it draws on various theories from the natural sciences, epidemiology, statistics, sociology, psychology and anthropology. Perhaps it was the existence of this diversity, the absence of a single, identified theory, which led Howie to his conclusion. A diversity that reflects the increasing complexity of health services research that requires different methodologies to effectively research the multiple phenomena that constitute health and health services provision.

The absence of a monolithic theory for general practice research is also a response to governmental restructuring of general practice. Methodological pluralism has been advocated by leaders in academic general practice as a response to rapid changes in the structure of health care delivery (Kinmonth 1995, Jones 1995, Olsen 1998). Seeking to improve individual health care, and simultaneously contain the rising cost of health care delivery, central government has sought to reform funding, organisation, management and clinical practice, as well as professional and undergraduate education. In this context, methodological diversity has been advocated in order to research and evaluate the benefits that policy changes claim for patients' health and for improved efficiency and economy in health care delivery. The inclusion of psychologists, sociologists, and anthropologists in academic departments of general practice has led to the introduction of methods from qualitative research (Kinmonth 1995, Jones 1995). For academic GPs like myself this has provided the opportunity to work with, and be supervised by researchers from different disciplines. I have learnt new research skills and used them to explore areas beyond the confines of the scientific method (see section 3.2.1).

The introduction of disciplines with differing philosophical and theoretical perspectives does, however, raise questions and tensions for academic GPs around what constitutes legitimate knowledge. The majority of research emerging from academic general practice is rooted in "scientism," presumably because of the nature of GPs pre-medical and medical education. According to Sorrell, scientism is the belief that the methods of natural science are the most valuable branches of learning. In this context, 'valuable' indicates the most authoritative, serious and

beneficial components of inquiry (Sorrell 1994). This situation immediately raises questions about the authority academic general practice attaches to qualitative methods and, specifically, the role qualitative methods have in creating the evidence and thus their impact in improving clinical practice.

3.2.1 Evidence and practice: The influence of *scientism*

Perhaps the most significant development to influence clinical practice and therefore the areas chosen for research, has been the drive to shift clinicians away from an opinion-based practice to practice based on reliable and valid evidence constructed through the application of empirical methods (Sackett, Haynes, Guyatt and Tugwell 1992). Known as *evidence based medicine* (Owen 1995) it draws on Cochrane's (1972) work which concluded that clinical practice and policy creation within the National Health Service should be controlled by science and, specifically, the randomised controlled trial. However, as commentators from within general practice have observed, Cochrane's ideas have had little impact on clinical general practice where epidemiological evidence has rarely translated into practice (O'Dowd and Wilson 1994). Explanations for this may include: a) epidemiological research performed outside general practice does not translate easily to the general practice context; b) epidemiological studies do not address the problems that most concern general practitioners; c) research has remained a minority activity carried out by a few practitioners mainly located in academic departments of general practice, producing research not easily comprehensible to GPs with little training in critical review of research and d) practitioners and researchers accept different forms of evidence, particularly when research evidence

conflicts with a practitioner's personal belief or experience (see example discussed below).

Previously, with a few notable exceptions (Fry 1966, Horder and Horder 1954), the majority of research evidence available to GPs originated from secondary care and was conducted by researchers not cognisant with the distinct nature, context and complexity of clinical general practice. For example, the diagnostic process, prescribing decisions and the interaction between doctors and patients during consultations, are multi-dimensional phenomena that involve not only recourse to "objective knowledge" but are imbued with, and affected by both individuals' beliefs, values and opinions of practitioner and patient. The consultation is a balance of revelation and concealment of thoughts and intentions practised by both patient and doctor, involving processes that can not be easily explained by quantitative approaches alone. This is evident in the work of those few GPs who have emphasised the value of the intuitive, narrative and interpretative aspects of general practice (Pendleton 1984, Pendleton and Schofield 1992, Neighbour 1987, Freeling and Harris1984, Heath 1995), which remain largely invisible to the scientific method but have been articulated through qualitative methods that capture social complexity.

3.2.2 Dilemmas raised by clinical practice: clinical experience vs. evidence

All GPs, and perhaps more so GPs actively involved in research, face the dilemma of balancing their scientistic beliefs and training with their experience of practising medicine in the community. In common with other clinicians, GPs use the

hypothetico-deductive model based on biophysical assumptions in making diagnoses (Ridsdale 1997). This has been defined as "the formulation, from the earliest clues about the patient, of a short list of potential diagnoses or actions followed by the performance of those clinical (history and physical examination) and para-clinical (laboratory, x-ray) manoeuvres that will best reduce the length of the list" (Sackett and Haynes et al 1992). In common with experimental research methods, clinicians generate working hypothesis (diagnoses) but instead of seeking information to falsify, clinicians have been observed to seek further symptom and signs to prove their hypothesis (Barrows, Norman, Neufield and Feightner 1982). Whilst this approach works well in the context of physical conditions e.g. myocardial infarction (heart attack) or hypothyroidism (under active thyroid gland), where precise pathological diagnosis may result in well delivered treatments leading to cure, it often fails when applied to patients presenting with symptoms related to the social, psychological and economic problems arising as a consequence of their experience of an illness (Howie1984, Morrell 1993). Morrell describes how GPs become confused and disillusioned when this occurs. To manage psychological and social symptoms, GPs acquired new skills and knowledge drawn from sociological, anthropological and psychological explanations of e.g. folk models of illness, the distinction between disease and illness, and doctor--patient roles and behaviour (Helman 1994, Pendleton1984). McWhinney (1983) has described the successful acquisition of psycho-social skills as akin to conversion to a new paradigm, "a very difficult but enlightening process." Thus, specific aspects of clinical practice challenge the universality of the hypothetico-deductive model that is based on the scientific method for managing all clinical presentations. My intention here is to illustrate how practitioners can draw

on their clinical experiences to understand why the scientific method is not always the most appropriate to research phenomena on general practice.

3.3 Locating the thesis

My research concurs with the view that the establishment of an evidenced-based medical culture within general practice will depend upon contributions from both qualitative and quantitative methods (Pope and Mayes 1993 and 1995, Kinmonth 1995, Jones 1995 and Britten 1993). In doing so I accept the proposition, that Richardson (1991) defines as post-modern, that there is ***"doubt that any one discourse has a privileged place, any one method or theory a universal claim and general claim to authoritative knowledge."*** In other words, this work acknowledges the necessity for methodological pluralism in studying the diverse and complex phenomena that constitute health care delivery in general practice—although acknowledging that the world does not tolerate all interpretations equally.

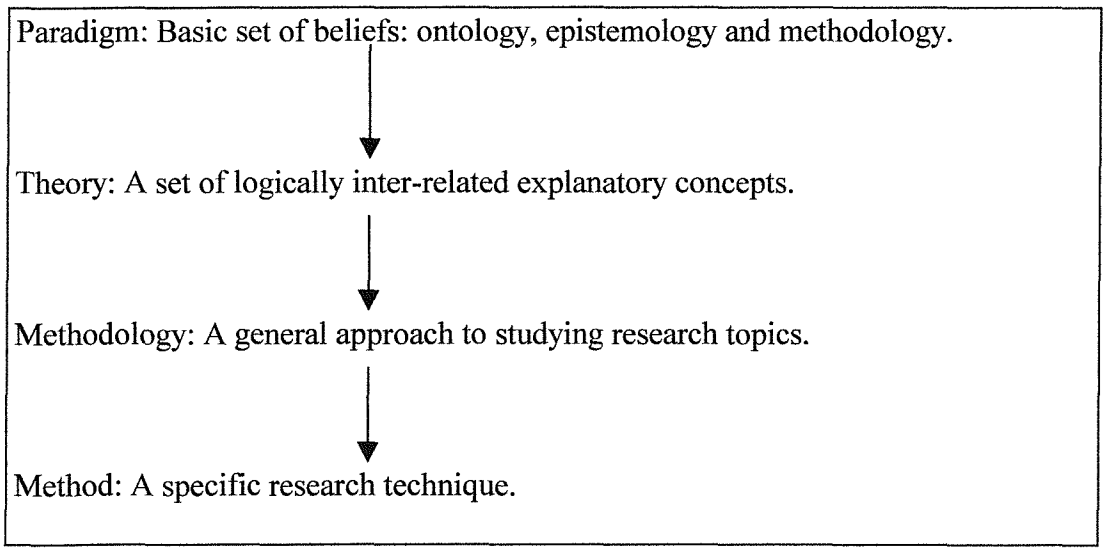
3.4 Rationale for using qualitative and quantitative methods

The decision to use qualitative and quantitative research methods was based on the theoretical needs of the research. My aim was to select methods that would provide the most valid and useful answers to the questions being asked. Ideally, the ways we ask questions (determined by our view of how knowledge should be constructed--epistemology), and the nature of the questions themselves (determined by how we construct our reality--ontology), condition the methods we use (methodology). Necessarily, how we ask questions shapes the nature of the

answers or knowledge created. The way questions are asked will be related to what we believe can be known about the research topic, whilst the nature of the questions dictate the form and media (words, numbers, diagrams, etc.) in which the data is assembled and then communicated to others (Kinmonth 1995). In this light, and in accordance with principles of qualitative research, I was guided by the nature of the questions with which I begin, rather than by an arbitrary commitment to a single methodological paradigm.

Qualitative research is committed to understanding and providing conceptual explanations of phenomena in context, whilst quantitative methods focus on the measurement and prediction of phenomena within an empirical framework. This thesis adopts the stance that the knowledge created by each approach is a legitimate form of evidence in its own right, the status of "evidence" being conferred by the intellectual authority each method gains from its own paradigm (Trusted 1997). The term *paradigm*, however, requires its own discussion, pursued below.

Box 3.1 The relationship between concepts used in research



3.5 Paradigms

The philosopher Thomas Kuhn has been most responsible for introducing the term "paradigm," into current intellectual discourse (Kuhn 1996). However, its meaning has remained contentious since its introduction in 1962. Kuhn himself is reported to have used it in at least 21 different ways (Masterman 1970). Masterman focused on one meaning; ***"a paradigm as a concrete example or procedure which entails a way of seeing."*** In the context of this work, I draw on Guba's (1990) definition which echoes Masterman's meaning: ***"a basic set of beliefs that guides an individual's action."*** For an individual or group, a paradigm is a worldview, defining the principles by which they make sense of the world around them. Examples include theologies (Christianity, Hinduism), cosmological belief systems (astrology) and legal systems, such as the adversarial system of justice. Paradigms deal with ultimate or first principles, granting them an aura of truth. Nonetheless, they should be regarded as human constructions. The *beliefs* are described as *basic* in the sense that they must be accepted on faith. Typically, paradigms are not subject to investigation through falsification but are assumed *a priori*. In this way, as indicated in figure 3.1, paradigms differ from theories. In the context of this thesis, quantitative research is part of a positivist paradigm, while qualitative research methods are united by a commitment to a naturalistic worldview. However, the labelling of qualitative research as a paradigm remains contentious for some contemporary qualitative researchers (Atkinson1995) because, as a site where different paradigms intersect, it is in some sense, a *meta-paradigm,* beyond the notion of a paradigm as an enclosed system. These different paradigms include phenomenology, ethnomethodology and symbolic interactionism (see section 3.6.3)

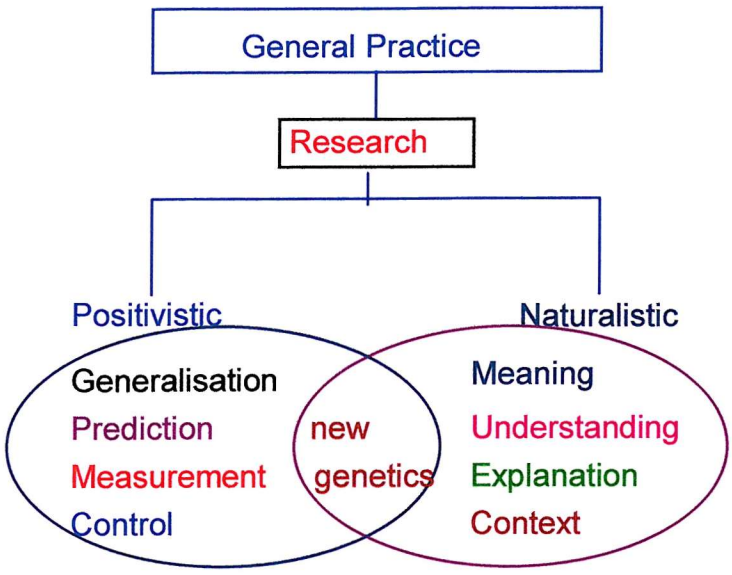
What unites qualitative research paradigms is their commitment to naturalistic enquiry and the fact that, in the context of general practice research, they are practised against the backdrop of the dominant positivistic paradigm. The terms positivism and naturalism are discussed later in this chapter.

3.5.1 Research and paradigms

In undertaking research the selection of an appropriate paradigm defines for the researcher the nature of the researched and delineates the limit of legitimate inquiry. In other words, the paradigm provides a framework for researchers, not only about the type of phenomena that can be studied and how they are to be researched but about how knowledge gained can be sensibly assembled. In this way, the paradigm conditions the nature and shape of the results that it could be seen to `discover.' Box 3.1 shows the relationship between the concept of paradigm to theory, methodology and method.

Insight in to a researcher's *basic beliefs* can be gained through answers they provide to some fundamental questions, see below. In designing and executing this research it was necessary for me to answer these questions. My answers reveal the basic beliefs underpinning my research and interpretations of the implications genetic advances have for general practitioners. In answering these questions, it became clear that this research like its subjects, genetic advances and general practice are sites where different paradigms intersect as illustrated by fig 3.1.

Fig 3.1 Genetic advances and general practice research



3.5.2 Defining paradigms; answering fundamental questions

a) Ontological Questions

What is the nature of reality and what is there that can be known about it? Or in other words, what kinds of things are there in the world that we can study?

In the context of discovering genes, a molecular reality exists which is amenable to exploration by using empirical techniques based on the scientific method. This leads to knowledge about the molecular structure and function of genes, as well as their impact on the cells, tissues and organs of the body. This knowledge adds to existing theories on heredity and disease aetiology. The application of this knowledge, e.g to predict, to asses individual risk, to develop treatments for specific human diseases such as cancer has implications beyond the microscopic. These include the ethical, psychological, social, legal, moral and philosophical

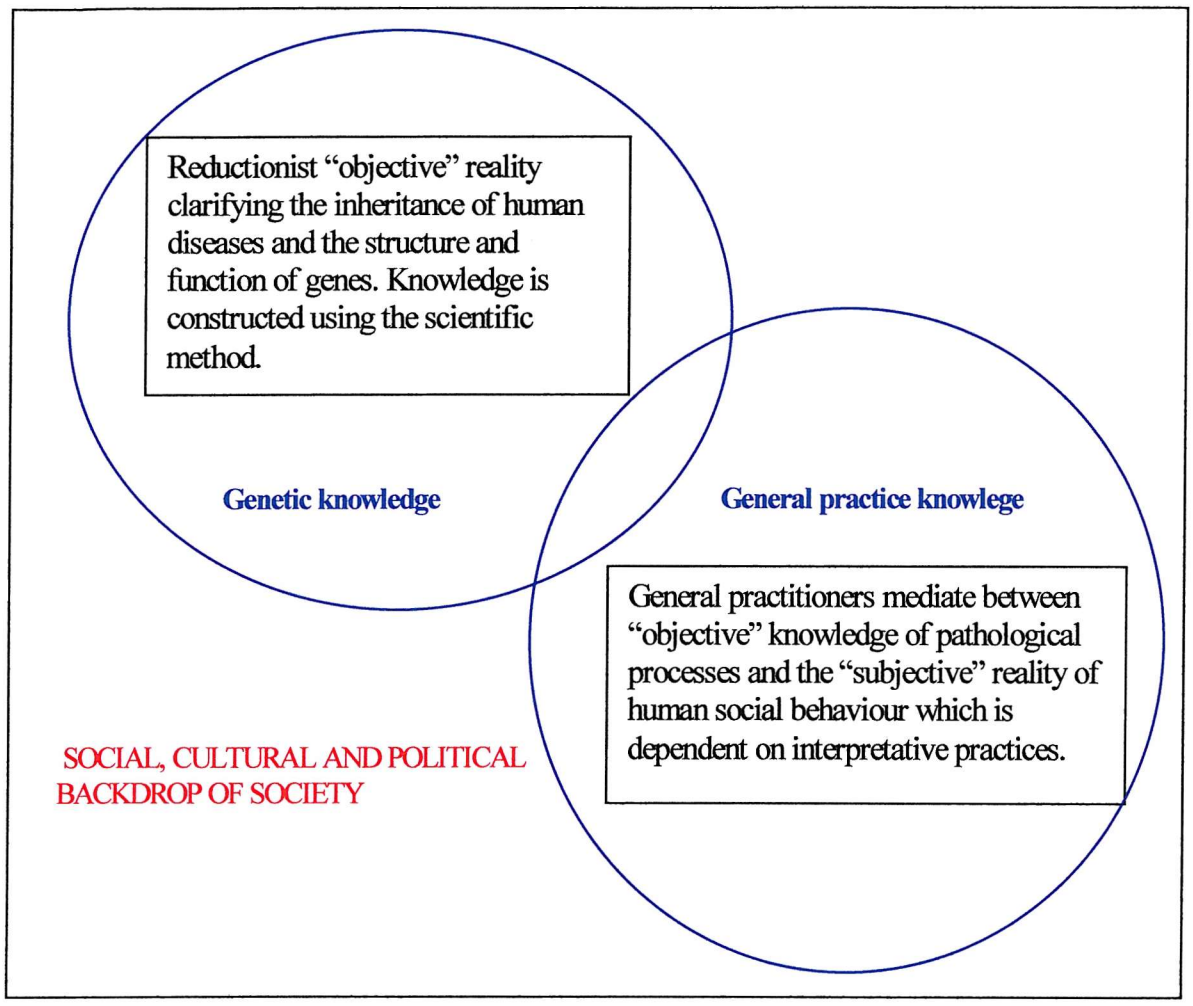
questions that arise as reductive knowledge interfaces with the interpretative and the subjective processes of human social behaviour.

Whilst the molecular scientist characteristically works with little attention to the political and social orders in creating knowledge, general practitioners occupy a position where "objective" knowledge intersects with the "subjective" world of social meaning constructed by interaction between people and their ideas. This is precisely the place where GPs can mediate between the molecular and the social realities of the new genetics as shown in figure 3.2. Thus, my research adopts a critical realist perspective that acknowledges the existence of many realities.

3.5.3. b-c) Epistemological and methodological considerations

These address questions such as; what is the character of our knowledge of the world? What is the relationship between the researcher and the researched? Briefly, epistemology is concerned with the philosophical claims about the way in which the world is known to us or can be made known to us and, as such, involves issues about the nature of knowledge itself.

Figure 3.2 Mediating between subjective and objective knowledge



Molecular knowledge about the existence, size, position, structure and function of cancer susceptibility genes e.g. BRCA1 and BRCA2, bolsters reductionist explanatory models of cancer aetiology in individuals and families. The value of this knowledge lies in its potential applications: the prediction of the existence and

position of other cancer susceptibility genes, the development of pharmacological interventions to control gene expression, and the development of predictive genetic tests to identify people at high risk of breast and ovarian cancer before the disease develops.

Knowledge is also required about how genetic advances are perceived and understood by GPs, for example: How do GPs think they can assess genetic risk for cancer? How do new genetic models fit with GPs' existing models of cancer aetiology? And what consequences may genetic advances have on GPs' duties and clinical behaviour?

Thus, methodologically this research echoes Denzin's (1970) assertion that, by combining multiple theories, methods and data sources we can attempt to overcome some of the intrinsic bias that comes from single-method, single-observer, and single theory studies (the relationship between the researcher and the researched is discussed in chapter 4).

3.6 The philosophical assumptions of qualitative and quantitative research

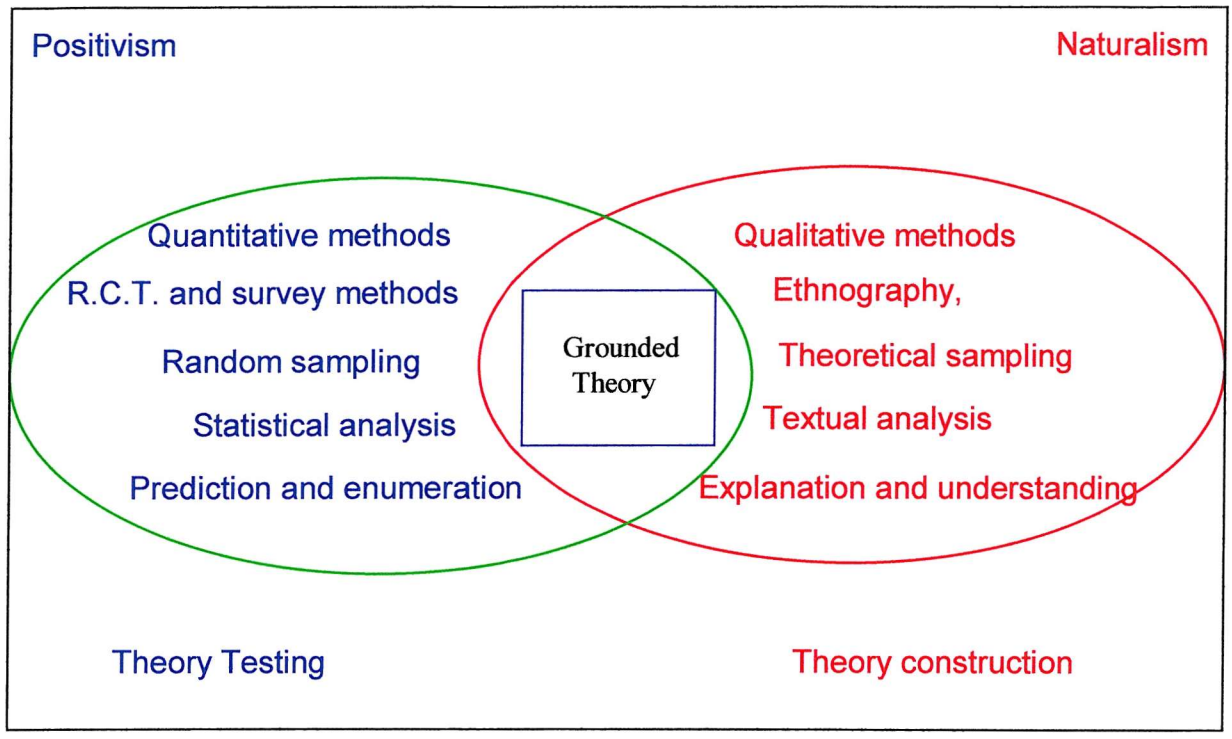
Earlier, I referred to the dominance of the scientistic view in general practice, which is derived through pre-eminence given to quantitative methods and the experimental model during premedical and medical training. Whilst it is not my aim to discuss in detail the philosophical claims of quantitative or qualitative research as competing models for researching general practice it is important to reflect on their differing stances. Guba and Lincoln (1982) refer to qualitative and

quantitative research as being founded upon divergent paradigms determined by their differing intellectual commitments to how knowledge about the social world can be constructed. Figure 3.3 depicts the often-polarised presentation of positivism and naturalism. Whilst this may accurately reflect the differences in their philosophical assumptions, the space between is linked by a spectrum of methods. These are generally categorised as qualitative or quantitative but there are methods that have attempted to occupy the middle ground. For example, "The Discovery of Grounded Theory" (Glaser and Strauss 1967) was published as general methodology applicable to quantitative studies as well as qualitative studies; *"we believe that each form of data is useful for both verification and generation of theory...in many instances both forms of data are necessary".* However, the text has had little impact on quantitative researchers. Subsequent publications; "Theoretical Sensitivity" (Glaser 1978) and "The Basics of Qualitative Research" (Strauss and Corbin 1990) have had their impact almost wholly on qualitative researchers. However there is increasing dissatisfaction with the failure of purely quantified results in certain clinical contexts, e.g. clinical recommendations based on the results of randomised controlled trials fail to produce results at the level of the individual (Pope and Mays 1995). In the context of my research, I moved from one framework to the other, I defined the relevant issues and constructed hypotheses using grounded theory methods and then developed a questionnaire to survey the opinions of a random sample of GPs.

In the next few sections, I shall reflect on the salient differences between the two approaches, the basis for doing this has been discussed in the introduction to this chapter. Knowledge of both philosophies was useful because it clarified how each

approach can research the same topic from different perspectives. The result, the combined outcomes of both approaches has the potential to provide a more complete picture of the researched topic than if either model is applied in isolation. It does not automatically follow that the validity of the final picture has to be greater than that of the individual studies. In other words, the validity of the final picture is not simply a sum of the quantitative and qualitative studies in this project. The idea that data produced by different methods from different paradigms can be unified to produce "one truth" is not without its problems. Hammersley and Atkinson (1993) argue that data can only be understood in relation to the purposes for which they are created e.g. for the production or testing of theory. If the purposes differ then they can not be integrated. I only wish to recognise this dilemma here as an important issue for researchers and develop it further in Chapter 7.

Figure 3.3 A representation of the space between positivism and naturalism



3.6.1 The positivist tradition

It is not my aim to give a comprehensive account of the development of positivistic thought but simply to indicate its roots and long and complex history in Western philosophy. The beginnings of positivism have been traditionally traced to Aristotle's (384-322 BC) view of empiricism as the foundation of human knowledge (Trusted 1997). The more proximate origins of positivism lie with Bacon (1561-1626) and Descartes (1596-1650), both men were concerned with creating a framework of thought and action that would provide certainty about knowledge of the world (Gower 1997). Bacon, unlike Descartes, argued for the authority of experience, experiment and induction. He emphasised a theory of

knowledge based on the methodical accumulation of experientially tested knowledge. Descartes (1995), however, expounded the certainty of mathematics as the fundamental instrument of scientific knowledge. He believed that certain knowledge could be created by logical deduction from self-evident premises. The common ground between their diverging views was the quest for certain foundations of human knowledge. Although Bacon articulated the importance of the experimental approach it was Galileo who, in his work on motion, "Two New Sciences" (1633) employed experiments to persuade readers that his conclusions were correct. Galileo helped to develop the role of experiment, as a means of discovering and understanding the laws of the natural world. Galileo put into practice Bacon's philosophies and so laid the foundation for the scientific method. Gower (1997) asserts Bacon's philosophy was taken forward by Locke, Hume and the other empiricist philosophers who all gave pre-eminence to the experiment. According to Hughes (1996) Descarte's certainty of mathematics as embodying timeless and unchanging principles became the language for expressing empirical knowledge.

The view that society, including its value and beliefs could be studied using the scientific method, was first put forward by Comte in the early part of the nineteenth century. It was developed subsequently by Mills, Spencer and Durkheim to culminate in the *logical positivism* (Kolakowski 1993) of the 1930s and 1940s, which promoted the use of the experiment and the survey in researching human social actions. Here positivism revolved around a central assumption that the scientific framework could be applied to determine the nature of any form of knowledge. However, more recently, Giddens (1979) suggested the term

positivism has become one of opprobrium and critical attack both in sociology and philosophy with little consistency in its use. He describes several competing and overlapping strands of positivistic thought in philosophy. However in the context of this research the philosophy of positivism referred to is that of the positivistic tradition in sociology, which broadly depends on the assertion that the concepts and methods of science can be applied to form a "science of man" or a "natural science of society" (Armstrong 1996).

Positivism as it affects sociology and therefore those areas which overlap with general practice e.g. social basis of disease, the social experience of illness or the role and behaviour of GPs, may be summarised as follows:
Physical science, conceived in terms of the logic of the experiment, is the model for social research. Although the form of the scientific method varies across the physical sciences and within medicine, it is unified by the underlying logic of the experiment. In general practice this logic is found in the randomised controlled trial.

Box 3.2 The scientific method as applied to clinical trials (Pocock 1995).

Define the purpose of the trial	: state the specific hypothesis
Design the trial.	: a written protocol
Conduct the trial	: good organisation
Analysis of the trial	: descriptive statistics, tests of hypothesis
Draw conclusions	: publish results

The clinical trial design involves separating the variables of interest from their social context and entering them in to a controlled research environment and then trying to fit the results back into the original context.

Universal laws. Positivism adopts the "covering law" model to explain events which is provided by reference to universal relationships between variables which hold across all relevant circumstances. In the context of general practice and the randomised trial, sampling strategies and statistical models are used to ensure the relationship between variables have a high probability of applying across relevant circumstances. The aim is to achieve generalisability of findings.

Neutral observation language. Priority is given to phenomena that are directly observable e.g. the number of antibiotic prescriptions issued for patients attending with sore throats. Facts about phenomena are collected by means of methods, which like the "facts" they collect are regarded as "theory neutral", otherwise they could not provide a conclusive test of the theory (Kuhn 1996).

In the positivist tradition research findings are reported in the passive voice. This serves to emphasise the belief in objective knowledge. This tradition is followed by bio-medical journals and all research including qualitative research is still required to adopt this style. For qualitative researchers this style of reporting can be problematic, given their philosophical stance on objective knowledge as described below.

3.6.2 Naturalism and qualitative research

Lofland and Lofland (1984) noted that not all social scientists use the same labels in the same ways when talking or writing about qualitative research, consequently, there are a diversity of terms and concepts used to describe qualitative research methods. 'Naturalism' and 'Qualitative Research' are just two labels used to signify a field of inquiry that has developed in response to the rejection of the notion that social reality can be studied through the application of the natural sciences. This was summed up by Matza (1969) to "remain true to the nature of the phenomenon under study."

In this thesis, I shall use the term qualitative research instead of naturalism, as it is the most common label to appear in bio-medical journals. Qualitative research is a field of inquiry in its own right, which draws on the work of researchers from many disciplines including anthropology, sociology and education. As a site of research, it represents the point where theories and methods from disciplines which accept the naturalistic perspective intersect. Similar to general practice, qualitative research has no theory or paradigm that is distinctly its own, its history and methods are those of the many disciplines that contribute to its methodological base. As Denzin and Lincoln (1994) assert qualitative research cuts across traditions, disciplines, fields and subject matter.

3.6.3 Origins of qualitative research

Hamilton (1994) identifies Kant's "Critique of Pure Reason" (1781) as the ultimate root of qualitative thinking. Kant took on the task of resolving tensions between Descartes and his sceptics, specifically the tensions between those followers of Descartes who accepted the pre-eminence of mathematics and objectivity in securing reliable and truthful observations, and those who refuted Cartesian absolutism (See section 3.5.1). Kant broke with Cartesian objectivism by proposing that visual perception was more than seeing. Implicit in Kant's conception of human knowledge as the product of sense perception acted upon by mental manipulation, is an acknowledgement that human knowledge is arrived at under the influence of the observer's thought processes. This laid open the way for the development of qualitative epistemologies.

The epistemology of qualitative research is a contested and complex field as illustrated by Atkinson's (1995) comments on attempts to classify qualitative research by authors who write on qualitative research texts books:

"draw together issues and approaches of very different levels of generality Some represent theoretical schools or traditions that have some affinities with qualitative research (symbolic interactionism, ethnomethodology, phenomenology); some are labels for general approaches to research (ethnography) or strategies or research design and theory (grounded theory); yet other are of extreme generality and have no claim to research methods at all (constructionism, deconstructionism, feminism and critical theory)".

Keeping Atkinson's points in mind, I was sympathetic to Bryman's (1988) "five tenets" of qualitative research; phenomenology, symbolic interactionism, *verstehen*, naturalism and ethnogenics. These are vast fields in their own right with their own sets of philosophical assumptions about the nature of social reality and to describe them here would be a substantial undertaking. My aim here is to highlight that qualitative research as a field of inquiry is a continually contested site. My need to understand its philosophies and methods operated against this shifting background.

3.7 Defining qualitative research

Defining qualitative research is problematic given the different constituents. However, uniting factors are its contested nature, and the complexity of its different roots. The definition I have found useful comes from the *Handbook of Qualitative Research* (Denzin and Lincoln 1994). It is also the definition that has appeared most often in bio-medical journals.

3.7.1 A definition

" Qualitative research is multi-method in focus, involving an interpretative, naturalistic approach to its subject matter. This means that qualitative researchers study things in their natural setting, attempting to make sense of, or interpret, phenomena in terms of the meanings people bring to them."

This definition is useful because it makes explicit the differing commitments of qualitative and quantitative research. One of the first requirements of quantitative

research is a constancy to the logic of the scientific method. In qualitative research commitment to methodological principles is replaced by commitment to the phenomenon under study, attempts are made to study events in a near a natural state as possible in contrast to the simulated environment of the experiment.

The definition makes explicit the philosophical assumption that the nature of social phenomena are unlike physical phenomena, in that they are human constructions guided by intentions, beliefs, rules and values. Finally it acknowledges that the creation of knowledge using qualitative methods is interpretative and so influenced by researcher's political and social value-framework in contrast to the positivist stance which claims the researcher operates objectively outside their value-framework.

3.8 Tensions surrounding the use of qualitative methods in general practice

The decision to use both qualitative and quantitative research methods was based on the theoretical needs of the questions being asked and on the practical need to secure funding for the research. Although there is an increasing interest in the use of qualitative methods in general practice (Whittaker 1996) and deepening understanding of how qualitative research can contribute to and complement quantitative evidence in academic general practice, decisions guiding the deployment and utility of the former are often made through a positivist lens. The result is that qualitative research most often occurs at the margins of quantitative studies, assuming a supportive role; for example qualitative research may be used to inform the design of quantitative tools, form the exploratory phase of a quantitative

study, conferring greater meaning to and providing context for the results of randomised controlled trials. Whilst these are legitimate roles for qualitative research its marginalisation can be interpreted in other ways. Is it perhaps a lack of familiarity with its it theories and methods that hinders more substantive deployment? Or are there issues of power at stake in the scientistic demand that qualitative research should speak not with its own fluency but to "us" in "our" terms? These questions relate to the status accorded to qualitative evidence by an essentially positivist discipline. In research which uses both qualitative and quantitative methods, the qualitative component becomes in danger of being driven by practical and theoretical needs of the quantitative project. Within bio-medicine, then, it appears that the intellectual authority of qualitative research, and so its potential to attract financial backing, are dependent upon a potentially compromising link to quantitative work. These tensions formed the backdrop to the design and conduct of this research.

3.9 The researcher in qualitative research

Qualitative research requires the researcher to be explicit about their personal and professional perspectives and the influence this has on data collection and analyses. Forming the backdrop to the design and execution of any study is the personal biography of the gendered researcher, who speaks from a particular class, professional, racial and cultural and ethnic community perspective. The gendered multicuturally situated researcher approaches the world with a set of ideas, a framework (theory, ontology) that specifies a set of questions (epistemology) that are then examined (methodology and analysis). Every researcher speaks from

within a distinct community, which shapes, in its own way the research act and outcomes.

My pre-medical was dominated by mathematics and the natural sciences (physics and chemistry) and biology. A combination that juxtaposed the simulated environment of the laboratory with naturalistic observational approach of field biology. "Naturalism" (Matza 1969) is a term often used to describe the nature of social research and itself appeals to the model of the "naturalist" in biology.

Medical school continued the emphasis on the primacy of the scientific method counterbalanced by increasing clinical exposure. Clinical experience meant contact with the complexities of human intentions, beliefs, intersubjectivity, values and personal knowledge and ethics. These dimensions make the operationalisation of de-contextualised epidemiological results to the clinical encounter difficult. Some of the aims of qualitative research immediately resonated with aspects of my general practice training and my experience as a clinical general practitioner. General practice training emphasised the importance of the psycho-social dimensions to patients' experiences of illness and disease. It puts the onus on the professional to understand the patient's experiences, not just in bio-physical terms but in terms of the social and psychological contexts of their lives. In other words, general practice training, unlike many other aspects of medicine, promotes the acquisition and integration of new skills with the traditional skills of biophysical medicine.

3.9.1 Personal biography

Born in England I have lived and worked here most of my life. I have also worked as a doctor in Saudi Arabia and Nepal. As a British-Indian there have been two major cultural influences on me, Hinduism at home and Christianity at school and work. This has led not simply to a culturally bifocal perspective. Hinduism itself is pluralistic and multifarious in its concepts of spirituality, divinity and the purposes of existence, while, in my experience, Christianity, though in some sectors responsive to multiplicity, tends, at a theological level, to unitary interpretations. In this way, my concepts and constructions of Hinduism and Christianity, as components of my own world view, echo, and stimulate the integration of, distinctions between quantitative and qualitative research. Like the grounded theorist one makes sense of the competing social frame works by a process of constant comparison, negotiating new frameworks of meaning.

3.9.2.General practitioner as qualitative researcher

Qualitative research may be seen as a "*bricolage*," a collection of research strategies from multiple interpretative paradigms. The qualitative researcher has been described as a "*bricoleur*." A bricoleur is a *"Jack of all trades"* who produces a bricolage that is pieced together, a close-knit set of practices that provides solutions to a particular problem in a concrete way (Levi-Strauss 1966, Denzin and Lincoln 1994). A parallel may be drawn with the clinical role of the General Practitioner. A GP may be seen as the bricoleur of medicine. During a consultation the GP may access knowledge and information from several different specialities in

medicine, in addition to information gathered from the patient. The GP will be aware that the same phenomena, the patient's symptoms, could be understood and explained differently by medical practitioners from other specialities. Furthermore, a GP's gender, age, social class, race, ethnicity and personal belief systems, as well as those of the patient may further influence the final interpretation or diagnosis. Similarly, the qualitative researcher as bricoleur is aware of the many different paradigms that can be brought to any particular problem, and understands that research is an interactive process shaped by his or her beliefs, gender, age, social class, race and ethnicity, as well as those of the researched. The process of interpretation and the final diagnoses constructed in terms of physical, psychological and social aspects of illness may be taken to signal the existence of multiple dimensions of disease as a phenomenon.

3.10 Summary

In this chapter I reflected on the different philosophical and theoretical positions occupied by qualitative and quantitative research. For me, writing this chapter helped to clarify the boundaries of qualitative and quantitative research and what each type of research sought to achieve. I discussed the construction of knowledge in general practice and highlighted how experiences in clinical phenomena can help GPs understand the role and power of qualitative research. Writing this chapter contributed to my development of theoretical sensitivity and reflexivity which helped me to construct a more theorised interpretation of the interview data. In acknowledgement of my role as research instrument I reflected on my personal biography.

Chapter four

Qualitative method: grounded theory

4.1 Introduction

Chapters one and three identified the reasons for undertaking a qualitative study. In chapter three, the philosophical and theoretical issues raised by undertaking qualitative and quantitative research were considered, allowing me to reflect on theoretical underpinnings of qualitative and quantitative studies. In this chapter, grounded theory, which guided the GP-interview study is presented. Specific attention is given to why this particular method was selected and the current controversies surrounding its use. The chapter begins by clarifying the aims and objectives of the qualitative study, then describes grounded theory methodology and how the grounded theory, interview study of general practitioners was undertaken.

4.2 Aims and objectives of the study

The core aim of the qualitative study was to explore the impact of genetic advances, and in particular breast cancer genetics, on general practitioners' perceptions of their clinical roles and professional identity. This was achieved through addressing a number of broader aims, some of which were identified during data collection and analysis.

4.2.2 The broader aims

The broader aims of the study were to understand and provide a conceptual explanation of general practitioners' perceptions, beliefs, concerns and experiences of genetics within the context of their routine clinical practice. Additionally, to understand and provide a conceptual explanation of general practitioners' perceptions, beliefs, concerns and experience of how recent advances in breast cancer genetics may affect their clinical practice. From these broader aims developed more focused aims:

1. To explore how the discovery of genes for breast cancer affected GPs' collection and use of patients' family histories.

2. To understand GPs use of the terms "familial" and "hereditary" disease, a theoretical aim that emerged from the analysis of GP interview data, and discussed in chapter five.

3. To give voice to GPs opinions and concerns in a field where the GPs' roles were being shaped by those outside of general practice.

4. To contribute to the debate between GPs, policy makers and specialists.

4.2.3 The objectives

From these aims a number of specific objectives were derived which are enumerated below:

1) To identify a group of GPs who had information and knowledge about the recent advances in genetics.
2) To understand how GPs currently conceptualise genetic disease.
3) To explore GPs' perceptions of their genetic work load.
4) To explore GPs' current strategies for discussing genetic risk in the context of established Mendelian disorders (e.g. cystic fibrosis) and chromosomal disorders (e.g. Downs Syndrome).
5) To explore how GPs make sense of a family history of breast cancer.
6) To explore GPs' perception and understanding of how heredity and the environment may influence cancer aetiology.
7) To understand if, when and why GPs collect family history.
8) To explore if and how GPs use family history as a genetic tool to assess risk for breast cancer.
9) To explore GPs' attitudes to providing genetic counselling and information for screening services.
10) To provide information for the development of a closed questionnaire for the GP survey study.

4.3 Grounded theory methodology

There are a range of qualitative strategies for data collection and analysis e.g. ethnography, phenomenology and grounded theory (Denzin and Lincoln 1994, Pope and Mayes 1993 and1995) many of which could have provided a suitable framework. However, grounded theory (Glaser and Strauss 1967, Glaser 1978, Strauss and Corbin 1990) was selected because it possesses a number of specific features; it was specifically aimed at hypothesis construction, which was useful in the context of this study as there was little published research. Grounded theory offered a systematic and explicit framework for sampling, data collection and analysis, with clear guidelines for how a grounded theory could be judged. These features made it understood by and so accessible to a broader audience. In medicine, examples of grounded theory research had appeared in bio-medical journals (Green 1993, Kai 1996, and Kai 1996), and so (in comparison to other qualitative methods) it was more likely to be familiar to medical audiences, and specifically to general practitioners. Another important attribute of grounded theory is the importance placed on iterative data collection and analysis, which affords opportunities to test the integrity of the data and its analysis. This in turn has the potential to maximise internal and external validity, processes regarded in biomedicine as important if this knowledge was to be considered as evidence. Finally, because grounded theory builds hypotheses from data and because it researches process it has the potential to close the gap between research and practice-- an important feature given the acknowledged costs to the NHS because of the lack of research in to process (MRC 2000).

4.3.1 "Grounded Theory" or "Full Conceptual Description"

Grounded theory was first described by Barney Glaser and Anselm Strauss in "The discovery of grounded theory, strategies for qualitative inquiry" published as a book in 1967. The method's foundations lie in the symbolic interactionist tradition which, according to Blumer, provided a framework for theorising the assumed complexities of human social interaction (1969):

"Human beings act toward things based on the meaning the things have for them; meanings of such things are derived from the social interaction that the individual has with his fellows; and meanings are handled in, and modified through an interpretative process and by the person dealing with the things they encounter."

Before the articulation of grounded theory, qualitative researchers would draw on such assumptions to inform data collection, analysis and interpretation. However, the exact processes involved at various stages e.g. making links within and between data sets to generate theory, or how sampling decisions related to the emerging analysis were not always explicit. This lack of transparency led to some researchers from social sciences and "non-interpretative" disciplines to label qualitative research as unsystematic, impressionistic, and lacking in rigour and credibility. Glaser and Strauss consciously set out to redress these criticisms by providing a reasoned account of the analytical

process that was explicit about how theory was developed during qualitative analysis—this was grounded theory.

Since the publication of "The discovery of grounded theory" (Glaser and Strauss 1967), the method has become contentious as is illustrated by the debate among qualitative researchers, and perhaps more significantly its co-founders, Glaser and Strauss. The crux of the debate centres on Strauss's challenge, in the guise of his publication (with Corbin) *Basics of Qualitative Research,* to the original description of the grounded theory method which I discuss below.

Following the original publication, each author has independently elaborated upon grounded theory, (Glaser 1978, Strauss 1987, Glaser 1992) and subsequently Strauss with Juliet Corbin in 1990. Other researchers (e.g. Charmaz 1983a, Melia 1997, Murphy et al 1998) have contributed to the debate between its co-founders. Specific points of contention have arisen because the original version was not explicit about the necessity of all the procedural steps, nor how they should be executed. For example, the role of saturation, the intention of progressive theoretical sampling, and how core categories were to be identified during data analysis. As Melia (1997) asserts, *Basics of Qualitative Research* (Strauss and Corbin, 1990) helped in "*laying out*" passages in the original, "*that are near mystical.*" Indeed Strauss and Corbin's version is noted for its detailed technical description, which presents the method at it most formulaic yet. It is paradoxical that "*Basics of Qualitative Research*" has created a methodological rift between Glaser and Strauss, whilst increasing grounded theory's accessibility and

acceptance in disciplines beyond sociology e.g. to nursing, general practice and education. It may be the procedural emphasis in Strauss and Corbin's version speaks directly to researchers who belong to applied and rule-governed disciplines, qualities which they bring to their research also.

Glaser and Strauss disagreed on the essential nature of the analytical strategy, the constant comparative method (Glaser 1992, Melia 1997). Glaser claims that Strauss and Corbin's version subverts the original aim to discover novel hypotheses grounded in observational data. In his opinion, the formulaic approach encourages researchers to identify categories prematurely and then collect data to confirm them. In other words, it joins other methods that privilege confirmation/ refutation over theory discovery or construction. Strauss and Corbin's version does have greater procedural emphasis which may be interpreted by some researchers as *methodolatory* (Janeseck 1994): the privileging of method over and above any other consideration. In this light Glaser describes Strauss and Corbin's method as *"full conceptual description...an almost new method borrowing an older name--Grounded Theory."*

A significant departure made by Strauss and Corbin from the original is the role of *saturation* -- the collection of data until a conceptual category becomes credible. Morse (1995) has defined pursual of saturation as the key to excellent qualitative work. This is because it promotes the collection of data to explore the dimensions of a specific category. For example, in this study I explored the categories of "familial disease" and "hereditary disease" by asking GPs and clinical cancer geneticists to give

examples of when and how these terms were used during consultations with patients and other health professionals. The respondents answered by describing the types of diseases they included under each of the labels and the context in which they were likely to encounter "hereditary" or "familial" diseases e.g. regular surgery sessions, ante-natal clinics, child health surveillance clinics and so on. In doing so they defined the dimensions of these categories which I then researched further by interviewing theoretically selected respondents e.g. interviewing the professor of genetics, a consultant geneticist and a GP with a special interest in genetics. In this way I was able to collect more data that increased the degree to which initial categories were saturated. Additionally, this process facilitated making links between the data sets to produce an explanatory narrative that connected data, which at the beginning had appeared diverse and unconnected. Thus pursual of saturation in combination with careful theoretical sampling has the potential to generate conceptual density and thus robust theorisation.

In the *Basics of Qualitative Research,* the emphasis on saturation is much less than on increasing *theoretical sensitivity* and the deployment of careful *theoretical sampling*--systematically gathering data relating to categories so that they are *theoretically proven.* The term theoretically proven indicates that certain concepts are deemed significant because they are repeatedly present or notably absent. In the context of qualitative research in primary care, increasing theoretical sensitivity and using careful theoretical sampling may be more achievable than the pursual of saturation. This is because, within the constraints imposed by bio-medical funding and the time required for data collection and analysis, saturation may not always a feasible option. Indeed

the question arises whether saturation is achievable in the context of primary care research. The term is seldom referred to in qualitative research published in bio-medical journals (Hoddinott and Pill 1997). This may reflect the current descriptive emphasis of much of the qualitative research emerging from primary care. In many instances, identifying simple categories for subsequent use in quantitative studies continues to be privileged over developing conceptual density and theorising the data.

4.3.2 Grounded theory contested

The defining constituents of grounded theory method will remain contentious as long as researchers who use this method continue to be critical and questioning of its procedural steps. This can be beneficial because of its potential to develop the method through clarification and refinement. Perhaps of greater concern is Glaser's move to silence any deviation from the original method. By essentialising grounded theory procedures he threatens to prevent further methodological development e.g. Shatzman's (1991) *dimensional analysis*. At best, Glaser's comments serve as a reminder of the pitfall qualitative researchers face in pre-judging categories which can lead to uncritical data collection and forcing data into ill-defined categories, which jeopardises discovery. Taken in this light, his comments simply reinforce principles of good qualitative research.

In conducting this study I have utilised both the original and the Strauss and Corbin versions of grounded theory. Thus pursual of saturation, careful theoretical sampling

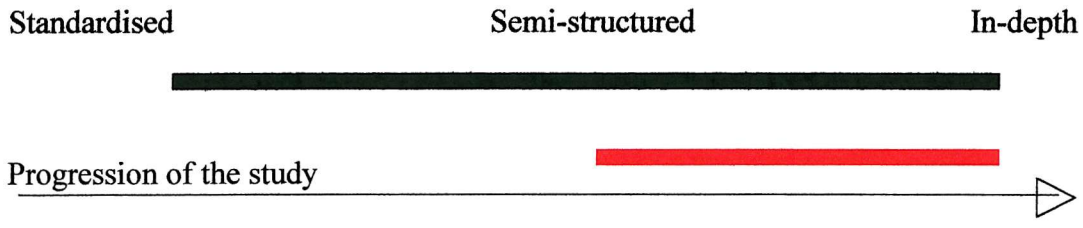
and increasing theoretical sensitivity were all used to maximise the construction and interpretation of categories.

4.4 Qualitative interviews

Once I had selected grounded theory, interviews became my prime method of data collection. Interviews have commonly been categorised as unstructured or structured (Burgess 1982b; Habermann-Little 1991, Fontanna and Frey 1994). This division is not entirely satisfactory, as no human interaction is without structure. Perhaps the more salient issue in qualitative interviews is the management of structuring during an interview. I found the degree to which my interviews with GPs were structured varied between interviews. In part this was determined by factors such as the respondent's or my ability to articulate ideas, the interviewee's anxiety levels, and the pressures on the GP's time. But other factors influenced the degree of structure too. For example, aspects of data and data analysis I needed to explore further became clearer as data collection progressed. Thus the completion and analysis of one interview automatically generated structure for the next by generating categories that required further exploration and testing. However, I had to balance my need to explore existing data and its analysis with encouraging GPs to speak un-interrupted about their opinions which had the potential for identifying novel and unforeseen insights. I was able to do both during interviews because I was prepared to be flexible, and sensitive to respondents' agendas. Here my perception of what had been said or withheld by the respondent played an important influence on the structure imposed.

Britten (1995) used the term "semi-structured" to describe interviews where structure materialises during the interview in response to respondent's narratives. McCracken (1988) has used the term "the long interview" to indicate a more flexible, responsive and detailed exploration. Unlike structured interviews, semi-structured and in-depth interviews are more flexible and sensitive to the interviewee's responses, which allows the researcher to explore the phenomenon in the context of the respondent's experiences. In figure 4.1 I have represented interviewing as a continuum and the red line characterises my perception of where my interviews lie. However it is important to point out that this is only an indication as the qualitative interview is dynamic and will have points where it is highly structured and other when it is freer.

Figure 4.1 The continuum between standardised and in-depth interviews.



4.5 An alternative to interviews

I could have collected the data using a series of focus groups. In primary care research focus groups have been advocated where the aim is to establish quickly the range of perspectives on an issue of importance among particular groups (Barbour1995). According to Kitzinger (1994) they can be used successfully for exploring group interactions and people's knowledge and

experiences, for examining not only what people think but how they think and why they think that way. This in-depth exploration using focus groups requires a trained or very experienced and skilled group moderator or facilitator who can facilitate discussions without biasing the discussion (Fitzpatrick and Boulton 1994).

I decided focus groups were not a feasible strategy for the following reasons:

a) Organising a group of general practitioners to attend at a specific time and venue would be difficult. GPs are constrained by their clinical commitments during the day e.g. patient contact in surgery, followed by home visits and the provision of emergency cover at all times during the day. To co-ordinate a group of 6 to 10 GPs all to be available for at least two hours without interruption would be difficult.

b) Duration of group from 1 to 2 hours; this would have required GPs to attend after finishing their clinical work which may have resulted in a low turn out.

c) GPs may have been inhibited from expressing their unfamiliarity about some of the issues raised by genetic advances in front of their peers.

d) I would have required training in facilitating and managing a focus group, which was constrained by finance and time.

Another potential approach could have been participant observation which was used to develop the initial interview guide. However, being present at consultations in which GPs' discussed genetic conditions or the genetic implications of a family history of cancer with patients and families would not have been a feasible option. This is because such discussions occur infrequently and are on the whole unpredictable.

4.6 Purposive sampling

Two main considerations guided selecting informants; the subject area of the new genetics, and the aims and objectives of the research. In this study, I used purposive sampling (Patton 1960). The logic and power behind this lies in the selection of respondents considered to be *information rich* because of their experiences of, or relation to, the phenomenon being researched. The assumption being that a great deal more will be learnt by asking questions of information rich respondents rather than a randomly selected group of people. However, to realise the potential for information richness the respondents and the researchers must possess an ability to articulate ideas clearly and be reflexive when talking about the phenomenon under research. Securing "rich information," increases the likelihood of conceptual density and rich description of the data during the analytical phases (Patton 1960). The overriding aim is not to secure representativeness in the sense of statistical random sampling. Unlike quantitative research, there are no published guidelines or tests of adequacy for estimating the sample size required for developing a grounded theory. The precise sample size is only known at the end of a grounded theory study (Morse 1995).

4.6.1 Identifying an *information rich* sample

I needed to interview GPs knowledgeable about genetic advances and specifically cancer genetics. Given the rapid pace of genetic discoveries it was unlikely most GPs would have kept abreast of developments. My search for informed GPs was resolved when the Wessex Faculty of the Royal College of General Practitioners organised a five-week course in September-

October 1995 titled, "Genetics for General Practitioners." Its aim was to introduce GPs to the scientific, clinical, ethical and societal issues raised by genetic advances. The course consisted of a series of 5 three hour seminars given by consultant geneticists, a clinical cancer geneticist and a genetic nurse specialist (the course details are summarised in table 4.1). Thus, the GPs who attended this course were a potential purposive sample.

Table 4.1 Topics covered by the genetics for GPs course

Basic Genetics e.g. modes of inheriting genes, linkage studies,
Recent developments in genetics e.g. discovery of the breast cancer gene
Screening and populations aspects of medical genetics e.g. cystic fibrosis
Cancer genetics e.g. identifying patients at risk of breast, ovarian and colon cancer
Clinical genetics in practice e.g. ethics of screening

4.6.2 Negotiating access to GPs

One strategy for gaining access to the GPs attending the course would have been to join the course myself. Although I could have approached individual GPs at the end of the course, I was more likely to secure access if I had been part of their group. However, on application I found the course was full and no allowances for extras could be made. I adopted another strategy, which involved volunteering to organise a general practice career booth (for the Wessex Faculty of the RCGP). This provided an opportunity to open-up a channel of communication with the Faculty course organiser who controlled access to the genetics for GPs

course. Once I had established a rapport with the course organiser I was able to re-present my case for attending the course. I negotiated attending two of the six sessions; *screening and population aspects of genetics*, and *cancer genetics*. Additionally, I was given permission to highlight my research and invite the thirty GPs attending the course to participate.

The two sessions I attending also proved useful for making specific observations e.g. the questions asked by GPs, the information the sessions presented, and hearing GPs' informal reactions to the sessions. This data together with the observations made by Dr Gantley (one of my supervisors and who attended the whole course) informed the construction of the initial interview guide. By the end of the course, ten GPs agreed to be interviewed and a further eight were interested but were unable to commit until they could review their existing commitments.

4.7 Making contact with GPs

As a practising GP, I was aware of the times when GPs would be available to speak to me. I gauged this would be either before or after home visits and before evening surgery. One of the significant benefits of being a GP-researcher was the ease with which I could contact and speak to other GPs. By introducing myself as a GP, I minimised any potential hindrances to speaking with GPs on the telephone.

I telephoned all 18 GPs who had expressed interest in the research two weeks after the end of the course. During the telephone conversation, I introduced myself, explained I was following up their interest in a research study. I explained what would be required of them; committing to

a date and time for interview, my need to audio-tape the interview, agreeing on a site for interview (always the GPs'surgery), and clarifying that I would require an hour of their time. Additionally, I stressed the confidentiality and anonymity of the interviews as well as the potential benefits of the research for GPs. For example, I emphasised the potential to voice to GPs' concerns about their future role in this area. Thus, having recruited all 18 of the GPs who had responded positively I decided to telephone the remaining 12. For this group I emphasised the research could providing GPs with an opportunity to have their say about some of the issues that had been raised during the *Genetics for GPs course.* This approach was reasonably successful because another six GPs agreed to be interviewed. In total 24 of the original 30 agreed. Of the six unable to take part, one was due to illness and the other five declined giving lack of time as their reason.

I then wrote to all 24 GPs enclosing a half-page summary of the study which detailed the broad aims of the research, the time commitment required, my own credentials as a GP and the anticipated benefits of the study, I also provided my contact details should the GPs have further concerns about being interviewed. I did this to reduce any anxieties GPs may have felt about the interview and to signal their control over the interview process. The key characteristics of GPs in the purposive sample are described in table 4.2 below.

Table 4.2 Key characteristics of the purposive sample

Purposive sample of GPs in face to face interviews	
Male	14
Female	10
Full time principals	20
Part time principals	4
Number with MRCGP	18
Overseas doctors	1
GPs in the sample who had other clinical commitments	
Clinical assistant in obstetrics	1
HIV counsellor	1

4.8 Development of an interview guide

Having selected the purposive sample and organised a timetable for interviews, the next step was to develop an interview guide. This involved drawing on observations made during the *Genetic for GPs* course (primarily by Dr Gantley who attended the majority of the course as well as from the two sessions I attended), insights gained from the literature review, and feedback from GPs who took part in the pilot interviews. The original interview guide is included as appendix one.

The interview guide was used for the first five interviews where it acted as an aide memoir ensuring I covered all the pertinent areas. After this, I found the interview guide hindered rather than aided data collection. This was for a number of reasons, firstly, my confidence in interviewing increased as time progressed, secondly, new issues identified by GPs during the interviews were explored in the next set of interviews and thirdly, I needed to test some of my interpretations of what had been said in previous interviews—see section 4.13.

4.9 Conducting interviews

Cornwell (1989) drew on Goffman (1959) to argue that in social interactions people are concerned with *managing appearances*. Cornwell argues that this is particularly salient in novel situations where one party feels unequal, and the situation feels awkward and unfamiliar. This is precisely when a respondent is likely to put on his or her *best face* and produce what Cornwell defined as *public accounts*. Such accounts represent a specific level of discourse where respondents prioritise giving opinions that they think the interviewer wants to hear whilst allowing the respondent to feel safe, and in control. These accounts are produced in response to the context of the respondent's situation and are not intentionally designed to mislead. My aim was to elicit careful and systematic descriptions of GPs' perspectives of their role in implementing advances in genetics, and specifically cancer genetics. I was committed to accessing GPs' own accounts of their understanding rather than the extent to which they conformed to existing opinions and policy statements. I wanted GPs' accounts to reflect, as faithfully as possible, the thinking with which they operated outside of the interview situation. To achieve these aims I needed a physical place to interview and a context where GPs would

not feel threatened, invaded, judged or unequal---perceptions, which were more likely to lead to *public accounts.* I needed the GPs to be relaxed, un-threatened and reflective, and to have a sense of equality, which I thought would produce more authentic accounts from GPs about their thinking.

However, it was possible for me to influence some of the factors that might have a negative impact on data collection. For example, I attempted to reduce respondents' anxieties about my identity and the interview process—the impact of doing so on the data collected was underlined in my first interview. I had assumed the GPs would have been clear about my identity in view of the number of contacts I made with them prior to interview e.g. during the *Genetics for GPs* course, during telephone conversations and finally through the letter confirming the interviews. However, some way in to the first interview I was surprised when the GP suddenly announced he had been trying to describe his thinking using words a social scientist would understand. The relevant extract of the interview appears below:

SK: *Can you to tell me about how you talk about risk to patients —for example in situations where well people are concerned about developing a common disease?*

GP: Is heart disease OK?

SK: *Yes- you might find it helpful if you draw on an example from practice?*

GP: ***Oh the area is so nebulous that you know damn well that nobody can calculate their risk. You give them information—a patient will accepts a certain amount of risk otherwise becomes life becomes socially inconvenient.***

SK Can you unpack that for me---what do you mean by socially inconvenient?

GP: Umm....what I mean is when I see some of my diabetic patients who smoke and you tell

them it is bad for them...umm I am trying to put what I'm thinking in to words that you'll understand—you're a sociologist are you?

SK: *Actually I'm a GP—I am a half-time principal at Aldermoor health centre and the rest of the time I'm doing this research.*

GP: **Oh—well that's different that makes it easier for me—you'll understand what its like!**

SK: You were telling about how you talk about risk to patients and you were saying something about social inconvenience.

GP: Well I am not very good at risk evaluation and the statistics of risk and probability—I'm good at statistics—I have an MSc and can do T-tests and Chi tests and what have you but probability I'm terrible with, for instance, I find it difficult to understand probability values for playing dice or cards. My brother who knows nothing about statistics, is wonderful at doing probabilities and can tell you exactly the odds of shaking a finite 2 or whatever it is. But then he is a gambler! It is the risk of probability I find very difficult to explain. Also I believe that I am basically ignorant and would not be giving terribly good information.

Explaining I was a half-time GP principal altered the GP's manner—he subsequently became friendlier, more ready to confide which is reflected in the content and pattern of his speech which became freer, more personal and open. GPs who have conducted studies involving face-to-face qualitative interviews have found emphasising their GP identity had positive effects on data collection even when the respondents were patients (Kai 1996, Hoddinott and Pill 1997). McCraken (1981) emphasises the need for the interviewer to reflect on how respondents may

categorise the researcher. Fontana and Frey (1994) underline the impact of being categorised by the respondent: *after one's presentational self is* ***cast*** *it leaves a profound impression on the respondents and has great influence on the success or failure of the task.* The GP in the example given above had made assumptions about my professional identity and affiliations, which influenced his openess in sharing information but also the nature of the language he used for doing this. Additionally, McCraken highlights that respondents may make similar assumptions according to how the aims of the project are presented, the dress and appearance of the researcher, and even the interviewer's pattern of speech. Thus my decisions about how I should present the project and myself could have a significant impact on the depth and credibility of the data collected. In general I decided to adopt a level of formality of dress, speech and presentation that reflected the professional environment in which general practitioners worked. This was not difficult to achieve given my own experiences of general practice. Below is a version of how I presented the research and myself. The content and emphasis of the message was altered according to the GP I was interviewing.

Thank you for agreeing to take part. As I explained in my letter I'm a half-time GP principal at Aldermoor Health Centre in Southampton and a researcher the rest of the time. I wanted to speak with you following the Genetics for GPs course you attended. I've recently received some funding from our local research and development office to look at the implications of genetic advances, and specifically cancer genetics for general practice. Because you've recently taken part in a course on genetic advances I would particularly like to hear your views. I'm especially interested to hear about your experiences of patients' inquiries about genetic conditions and how you manage them. You should feel free to talk about whatever you think is relevant and important to you and your patients. I'm not an

expert in this area and the interview isn't a test of your knowledge.

I am going tape record our conversation because it is an efficient way of my not forgetting what we will say today. I am going to analyse the data and hope to write a paper for publication. In the end I hope this work will voice views from general practitioners about our role and so influence those involved in shaping policy in this area. I will print quotes from some of the interviews but I will ensure your anonymity ---you won't be named and the exact location of your practice won't be revealed. Only a secretary and myself will have access to the tape recordings of your interview. I will send you a transcript of the interview for your comments. Are there any questions you would like to ask me about? Is what I've just said clear to you?

4.9.1 Interviewer as research instrument

In chapter three I reflected on my personal biography in relation to conducting qualitative interviews, here I discuss the concept further in relation to data collection. In qualitative research the researcher is his or her own research instrument. Data collection and analysis requires the researcher to use their experiences, imagination and intellect to create meaning out of their observations (Janeseck 1994). To be an effective instrument requires the researcher to be reflexive and transparent about their interaction with respondents, which in turn may help others in assessing the standards of the research. Within social sciences there is a considerable body of literature on *researcher as instrument* (Cassell 1977, Guba and Lincoln 1981 and McCraken 1988). However, the implications of this concept for research done in primary care is less well

developed (Richards and Elmslie 2000, Hoddinott and Pill 1997). Britten (1995) discussed *researchers as research instrument* in a methodological paper on qualitative interviews but limited her discussion to the more tangible and concrete aspects i.e. monitoring the directiveness of interviews, the time respondents were allowed to answer questions, and how to maintain control of the interview. She highlighted the pitfalls clinicians faced if they moved from being a clinician to an interviewer with the assumption that skills required for both were exactly same. To illustrate this Britten emphasised that no matter how patient-centred a consultation was, ultimately, it had to be re-fashioned and made sense of through biomedical frameworks, which allowed the doctor with the patient, to negotiate diagnostic and therapeutic decisions. Thus, she distanced the process of clinical consultations from qualitative interviewing, and so the skills and knowledge required for conducting consultations and qualitative interviews.

Mays and Pope (2000) emphasised the importance of demonstrating reflexivity and being explicit about how the researcher and the research process shaped data collection and analysis. For example, researchers need to consider how their gender, ethnicity, background, professional status, personal experiences, imagination, opinions and intellect impact on what is said during a qualitative interview and the analysis produced. Richards and Elmslie (2000), and Hoddinott and Pill (1997) are among the few who have engaged with these issues. Hoddinott and Pill considered the impact of a GP disclosing their professional identity on accessing and recruiting patients, and on the nature of the data collected during face-to-face interviews. They found definite advantages; improved access to respondents and improved data collection during

interview when the GP was explicit about their professional identity. However, they acknowledged that the topic of research, the research question and how closely it was associated with aspects of medical care would influence if the disclosure was beneficial. Richards (a GP) and Elmslie (sociologist) compared the impact of their professional background during face-to-face interviews. Their findings echoed those made by Hoddinott and Pill, but they found the category of *doctor* could overpower other personal characteristics of the researcher. For example, they found that Richards reported greater deference amongst working class respondents and social alignment among middleclass respondents. Richards and Elmslie categorised themselves as *white, female and middleclass* but they did not explain in detail how these characteristics influenced their interviews.

4.9.2 Maintaining distance

This section explains how I managed the tensions of researching my own professional culture. As a general practitioner I was already assimilated within the culture of general practice and its institutions. McCraken (1988) uses the term *manufacturing distance,* to signify a process where the researcher embarks on developing a critical awareness of their own assumptions and beliefs when they encounter familiar practices, symbols, or patterns of thinking which they might assume have meanings familiar to their own world. Whilst these assumptions may be apt in some instances at other times they could foreclose the range of issues identified and discussed as well as the depth to which they are explored. The concept of *manufacturing distance* or *making the familiar strange* comes from anthropology where investigators would

go and experience a different culture for an extended period of time. When they returned to their own culture they reported to have developed a profound sense of its peculiar and arbitrary nature. This route was not a practical solution for me—writing chapter 3, Shaping Knowledge, and access to a medical anthropologist (Dr Madeleine Gantley) facilitated the development of a critical distance. This reflexive preparation prior to interviews was vital for minimising the degree to which I might pre-define GPs' perspectives and remain open to new dimensions.

However, there were advantages for me to interview other GPs. For instance, I was more sensitive to GPs giving *public accounts* of their behaviour and I was able to respond by gently challenging and probing their responses in an attempt to access more of their own thinking. I also had an personal experience of the processes and phenomena they spoke of during interviews and so was able to assess the verisimilitude of their responses (Denzin and Lincoln 1994). However the disadvantage was that I could share the same "blind-spots" as other GPs and so would fail to probe or question taken-for-granted meanings and assumptions. This was minimised by reviewing interview transcripts with Dr Gantley with the aim of identifying points in the interview where the GP and I assumed we understood each other. In this way I was able to construct probes to explore meanings further—below are examples of some of the probes I used:

That's an interesting view –why do you think of x in those terms ? Other GPs I have interviewed put forward different ideas/arguments to the one you've just given me—what do you think about it/them? How do such views sit with your own ? Why do you think there is a difference? What have you based your thoughts on? Probes: professional/personal

experiences, evidence, policy statements?

This approach was also successful in encouraging GPs to reflect on their own opinions. It helped introduce a degree of critical distance for the respondent about their own assumptions. In other words, it created distance for GPs from their own taken-for-granted state. However, I realised the success of my challenges and probes to GP's depended on my vigilance of my own critical distance from general practice.

4.9.3 Site for interviews

Interviews were conducted at the GPs' surgeries in their consulting rooms. This had certain advantages: GPs were able to refer to computer and paper records of consultations with patients, they were able to access correspondence from consultant geneticists, and refer to family history records. Additionally, GPs were able to examine practice materials they had developed to collect family histories of common diseases. By referring to these materials GPs grounded their opinions of genetic advances in concrete clinical experience. They recalled consultations to challenge or support their specific responses. This was important as it increases the trustworthiness of the data because its origins are located in practice and not constructed entirely in the abstract (Hammersley and Atkinson 1993). It is important to emphasise here that GPs did not break with the principles of patient confidentiality at any time.

4.10 Data collection

Face-to-face, interviews were conducted with all respondents. To refine questions and my

interview technique I piloted the interview guide with six local GPs. These data were not included in the data analysis but feed back from these helped to fine-tune interview questions and my interview technique. The questions I asked addressed the following broad areas: GPs' opinions and beliefs about their role in providing genetic counselling and risk assessment; knowledge of advances in genetics; current skills they could immediately operationalise; current clinical situations where they provided advice on genetic risk; attitudes to extending current services to provide genetic counselling for common conditions, and biographical and demographic data. GPs moved from predictable statements of professional and practice policy to more critical reflection when I encouraged them to focus on a specific personal or professional experience. In order to seek respondent validation I regularly summarised and fed-back my interpretation to GPs during the interview. All interviews were audiotaped and transcribed verbatim. Respondents were offered a further opportunity to review the transcription as well as the final analysis. However all declined citing time constraints as their excuse. Data collection and analysis proceeded in batches of 5 interviews, which in keeping with grounded theory, guided decisions on further sampling—theoretical sampling (see table 4.2). Thus sampling, data collection and analysis were iterative.

4.11 Theoretical sampling

As data collection and analysis progressed I accumulated volumes of both data and analysis. However, as data analysis proceeded I was able to reduce the number of preliminary issues identified in the data by making links between them thus developing more complex analytical units: categories and concepts which umbrella larger volumes of data (see chapter 5).

However, I needed to assess the integrity of these units. The basic questions I asked as I analysed the data were: what groups or subgroups should I turn to next in data collection? And for what theoretical purpose should I selected them? I needed to collect data from respondents who were expert, articulate, critical, and able to provide, thoughtful reflection as well as rigorous criticism of the categories and concepts I had identified.

Thus the criteria for selecting constituents of the theoretical sample were those of theoretical purpose and relevance. My aim was to increase the depth, focus and integrity of the analysis. The aims of theoretical sampling have been summarised by Patton (1960):

1) to extend the data already collected,
2) to confirm/challenge the data and the emerging analysis,
3) to guide future sampling.

As data from the theoretical sample came in I was able to assess whether the links made between different data sets to establish the categories, concepts and themes were bone fide. This was possible because doing interviews in the theoretical sample phase I was able discuss the links and then explore if the respondents' confirmed or challenged my interpretations. Respondents in the theoretical sample were:

(F)=female, (M)=male.

1) A professor of general practice, involved in research, clinical practice and teaching (F)
2) An academic GP, half time principal and researcher. (F)
3) A full time, service practitioner who had not attended the course (M)

4) The director of a research network, trained in public health and general practice (F)

5) A service GP who had a specific interest in genetics but had not attended any genetics course (M)

6) A GP leader in genetics (M)

7) A consultant in cancer genetics (F)

8) Six GP principals from local practices, (3 F)

9) A GP teacher and tutor (F)

Table 4.3 The theoretical sample and specific issues explored

Theoretical sample, n=14	Specific issues explored
GP principal and professor of general practice	Hereditary and Familial conditions. Role of GPs. Generalism and specialism.
One lead GP and one advisor on health policy	Role of GP, generalism.
GP principal with specific interest in genetics	Role of the GP. Hereditary and familial.
GP teacher and GP tutor	Ethical information needs, holism and generalism
6 GP principals from local practices (no specific interest in genetics)	Impact of genetics on current practice, role of the GP, hereditary and familial conditions, generalism and specialism.
Consultant in cancer genetics	Familial and hereditary disease
Professor of genetics	Generalism and specialism

Members of the theoretical sample were identified from their published contributions to this area of research, membership of policy making and public committees considering the role of

GPs in this area and by the suggestions respondents made during interviews.

All interviews were done at a site convenient to respondent. All interviews were audio-taped and transcribed verbatim for analysis. As with the purposive sample all respondents were offered the opportunity to comment on the transcript and an initial analysis of it but all declined because of time constraints.

4.12 Assessing transferability

Finally, I fed back my analysis to a group of 20 GP principals from practices in Hampshire and Dorset. This was opportunistic sample of GPs who were attending a local research network seminar. The group consisted of service GPs, GP tutors and teachers as well as academics who were based mainly in urban and semi-rural practices. A summary of the data, final themes and analysis were presented to this group. They were asked to comment on how well they thought other GPs comments described their own perceptions and experiences with genetic conditions. By encouraging the assembled GPs to discuss our findings and interpretations we were able to establish the transferability of the analysis.

4.13 A *natural history* of the PhD

Appendix four is a time table representing the key events which occurred during the course of this work e.g. changes in supervision, extensions to my roles and responsibilities in the department, and changes to my contract and funding, all of which had an impact on the rate at which the research progressed. The maximum duration of a part-time thesis is six years, for me this period was between October 1995 to October 2001. However, for the last three years of this period the thesis was completed in my personal time. The period between October 2001 and April 2002 when the thesis was submitted was used to finalise the writing-up of the thesis and collate comments from Dr Gantley.

Changes in supervision. Dr Williamson and Dr Gantley ceased supervising the PhD in 1997 which was less than two years into the allotted time of six years. Dr Gantley was promoted to senior lecturer at a university in London and left in the first half of 1997. Dr Williamson left a few months following Dr Gantley's departure due to ill-health and stayed away from his post for eight months. Following their departures there was a period of four months when I was without a formal supervisor. This coincided with a stage in the research when I was moving from the qualitative to the quantitative study. The absence of a supervisor led to a delay in constructing the questionnaire from the qualitative data, testing the questionnaire and then executing the survey.

Acquiring a new supervisor was not straight forward because in addition to the loss of Dr Gantley and Dr Williamson, Professor Kinmonth, who could have assumed the role of supervisor was unable to supervise as she was due to leave for a chair at

Cambridge University. The only senior lecturer in the department who was left to supervise this PhD was Dr Helen Smith and she agreed to do so. Dr Smith and I negotiated with Dr Gantley that she act as an advisor to the PhD for the rest of its duration. My new supervisor was a quantitative researcher with little experience of qualitative research. Thus our first weeks were spent on re-examining the research, its methods, aims and objectives and my progress. The impact of all these events was to delay the development of the questionnaire and so the start of the quantitative study. The data collection for the qualitative study had been completed by the time Dr Gantley left and the analysis was in progress. I met with Dr Gantley once every two or three months following her departure to finalise the analysis and for advice on using the qualitative data to develop the questionnaire. All in all the change in supervisors resulted in setting the work back by at least six months.

4.14 Development of the aims and objectives of the qualitative study

Sections 4.2, 4.2.2 and 4.2.3 present a series of aims and objectives of the qualitative study. This presentation whilst useful does conceal the process of how some of the aims and objectives were identified from the interview data and its analysis. In this section I use the category of *family history* to illustrate how its further exploration through theoretical sampling helped define some of the key objectives.

The core aim of the study, which was defined at the outset, was developed from insights gained by attending the *Genetics for GPs* course and in response to the rhetoric adopted by experts, policy statements and the various people who were leading in this field e.g. Austoker, Harris and Harris, Quereshi and Calman as well

an statements from the RCGP (see chapters one and two). The core aim represented the general focus of the study. Expert opinion and policy documents advocated that GPs should systematically collect and record the family history of common cancers to assess an individual's and their family's genetic susceptibility to cancer. Policy makers regarded genetic risk assessment using family history as a pre-requisite to appropriate referral.

These insights were also used to construct the initial interview guide which was used in the first five interviews (see appendix one). The questions in the guide moved from exploring general to more specific issues. Initial questions focused on issues of structure and function e.g. on GPs' ideas of their roles in the context of genetic advances, how they understood their expertise and knowledge of genetics in relation to that of other secondary care professionals and what their experience of referring and managing patients with genetic conditions was. The first five interviewees were GPs in the purposive sample who were in a position to reflect on their experiences of attending the course and relate this to their professional context and practice. A recurring issue that emerged from the first five interviews was the use of family history in clinical practice and how this influenced what GPs defined as *familial* or *hereditary* disease.

Thus in section 4.2.2 which lists the aims, numbers one and four were identified at the outset informed by published literature. Aims 2 and 3 emerged from the analysis of the first five interviews.

Figure 4.2 Exploring family history by careful sampling

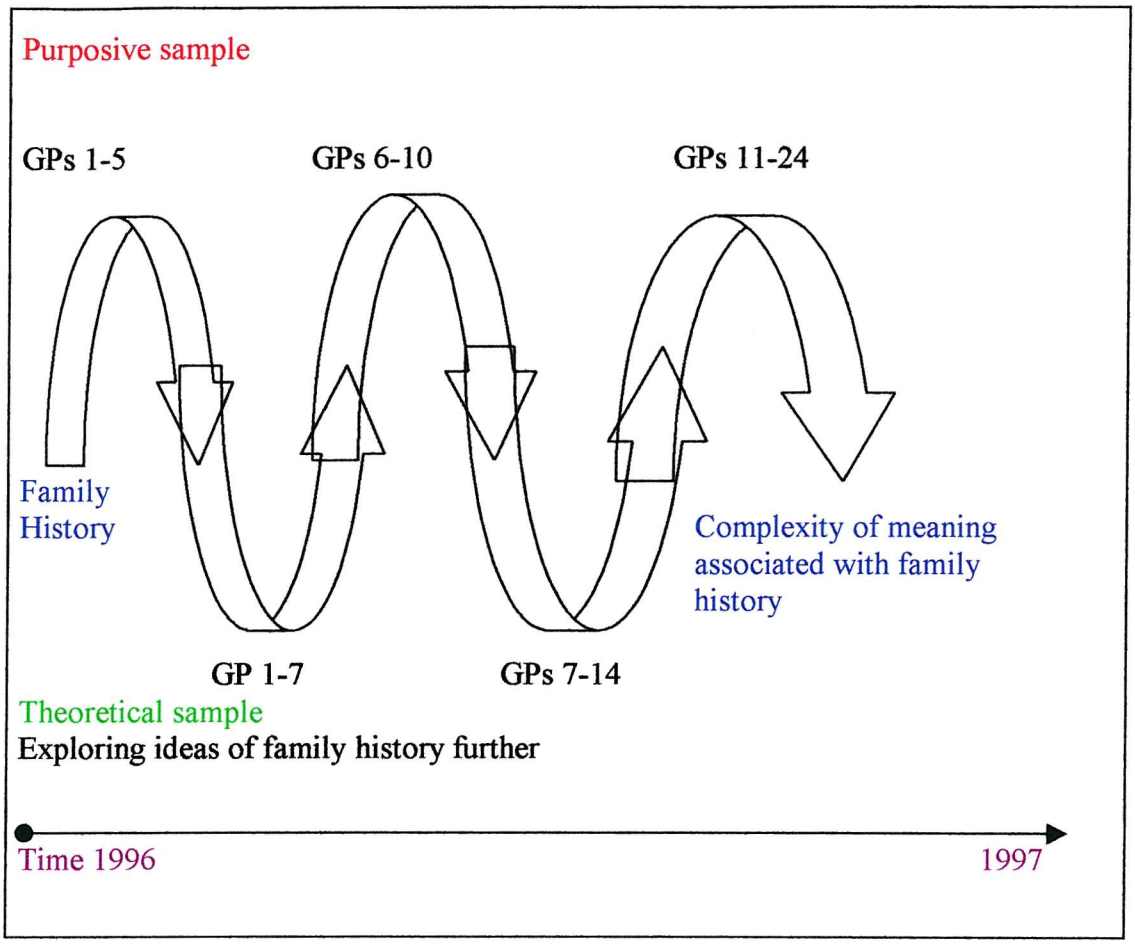


Figure 4.2 shows how the category of family history was explored with the five GPs in the purposive sample. They described how and why they collected family history in clinical practice, the contexts which triggered family history collection, and the meaning family history had in different clinical contexts (see chapter five theme one). These clarifications were then explored with GPs and experts who were selected as part of the theoretical sample in attempt to extend and the data. In this way ideas moved from the purposive sample to the theoretical sample and then back again to the purposive sample and so on. Each movement was mediated by me, the researcher and influenced by my interpretations of the data and by my

decision to focus on a specific part of the data. Decisions about focus were influenced by need to provide results that would respond to the assumptions of policy makers and experts. By the time I had interviewed the first seven respondents in the theoretical sample I had identified specific objectives. For example in section 4.2.3 objectives 2,3,4,5,6,7,and 8 emerged through this iterative process. The remainder of the interviews were used to explore these objectives as fully as possible.

4.15 Summary

In this chapter I have described the processes involved in deciding on the design of the study, identifying appropriate samples of GPs, negotiating access to GPs and collecting interview data. I also clarified how the aims and objectives were influenced by the iterative process of sampling, data collection and data analysis. In the next chapter I shall present the analysis of the interview data.

Chapter five

Qualitative analysis: process and results

5.1 Introduction

Having described how the data were collected, this chapter presents the analysis of the GP interviews. My general preparation for undertaking a grounded theory analysis was described in chapter threes and four, which underlined how interpretation in qualitative research is shaped by genre, narrative, stylistic, personal, cultural, and paradigmatic conventions. In this chapter I provide a more explicit account of the analytical procedure. The first section of the chapter describes the constant comparative method: how the raw interview data were coded in to issues, which in turn were linked to generate categories and concepts, which in turn were linked to produce the themes. The second section of the chapter presents the themes, categories and concepts with supporting interview data and its interpretation. Two themes are presented: genetics in the generalist context and implications of genetic advances for generalist identity.

5.2 Hierarchy of analytical units

There are a variety of terms used in qualitative research to describe different levels of analysis. Below is an explanation of the terminology I used and its significance in indicating the degree of abstraction achieved at each stage of analysis. The analytical units used were: issues, categories, concepts and themes—the order of the terms as

written reflects their increasing complexity and abstraction. Each theme encapsulates a range of categories and concepts which in turn were developed from a number of different issues identified in the interview data. Thus, themes embody a range of processes captured in the interview data and so represent the most complex and abstract units of interpretation.

5.3 Grounded theory analysis

Constant comparison analysis is the core analytical procedure in grounded theory. There are recognisable stages that make-up the analytical process: open coding, axial coding, and selective coding. I have summarised the procedure in diagram 5.1 below. The first stage in a grounded theory analysis is open coding: a process through which concepts and categories are identified and their properties and dimensions discovered in the data (Strauss and Corbin 1990). This involves reading the script sentence by sentence to identify initial categories—these may be key phrases or events described by interviewees which provide insight into the interviewees' (and sometimes the interviewer's) assumptions and practices. The initial categories and issues are then examined in terms of their properties and dimensions. At this stage the researcher reflects on the need to collect more data to explore further the properties and dimensions identified (see chapter four).

The next step is axial coding when the researcher reassembles the issues, and categories in new ways around a core category or concept categories identified during open coding. The aim here is to systematically develop new categories and discover

how categories relate to one another. It is during this phase of analysis, according to Strauss and Corbin (1990), that more analytical and theoretical ideas about the data begin to emerge.

The next step is selective coding, the process of integrating and refining the theory. The researcher identifies a "story" and writes a narrative that integrates all the categories identified in previous steps. In other words categories are organised around a central explanatory concept e.g. such as *the genetic window* described below. It is at this point a hypothesis may be constructed.

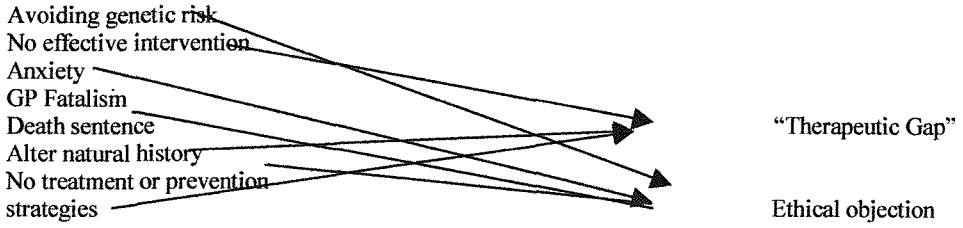
Figure 5.1 below, is a representation of the grounded theory analytical process—constant comparison analysis. It illustrates the preliminary moves and links that were made between data and categories to arrive at themes and then a narrative grounded theory analysis. The diagram is a schematic representation of the analytical process and its primary objective is to illustrate how data from different interviews were deconstructed and then reassembled. Thus it should be read as a basic skeleton of a grounded theory analysis. What a diagram can not represent is the complexity of how the researcher's theoretical sensitivity and reflexivity influence the research and analytical process. In reality the moves shown occur in a matrix where data, and ideas from literature, experience, imagination, reflexivity and theoretical sensitivity act together to produce the analysis.

Figure 5.1: Representation of grounded theory analysis:
Step 1: Open coding

Extract from Interview 1	Initial categories	Extract from Interview 5	Initial categories
SK: When do you raise the : issue of genetic risk?		SK: When do you raise the issue of genetic risk?	
GP: *I don't raise the issue-not for breast and ovarian cancer-because there is nothing positive that can be done at present. Raising the issue would only make the patient and the family anxious.*	Avoiding genetic risk No effective intervention Awareness and anxiety	GP: *The problem with breast cancer - what can you do if you've got the gene? Is there any sense in giving a potential death sentence? If at the moment we can't alter the natural history then I can't see myself raising the issue*	GP Fatalism Death sentence Alter history No treatment or prevention.

Open coding clarified questions to be asked in subsequent interviews e.g. Can you describe situations where you may be compelled to discuss genetic risk for breast cancer?

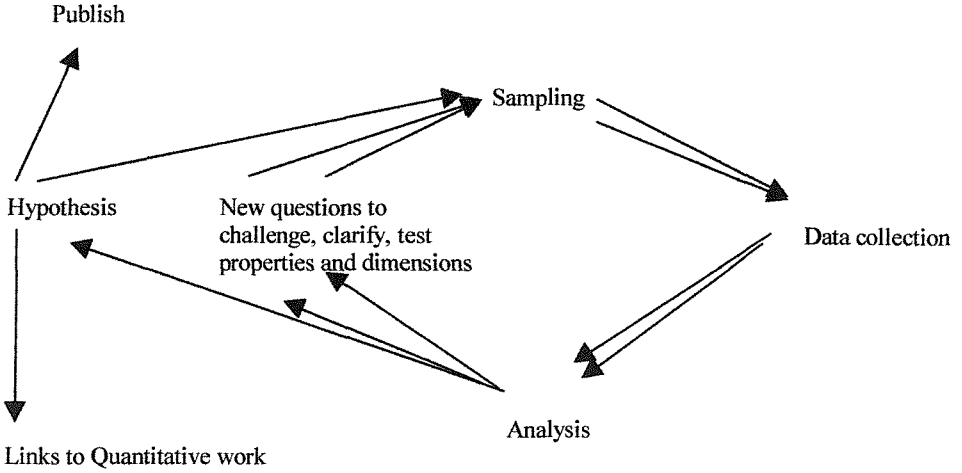
Step 2: Axial coding **Step 3: Core Categories**



Step 4: Selective coding: "Identifying a story."

Ethical dilemmas associated with the therapeutic gap: "I don't raise the issue-because there is nothing positive that can be done..."
GPs were reluctant to alert patients to genetic risk in the absence of effective screening technologies and therapies to reduce risk or prevent disease—in other words they identified a 'therapeutic gap' as an ethical dilemma. By raising the issue of genetic risk GPs thought they would create anxiety.

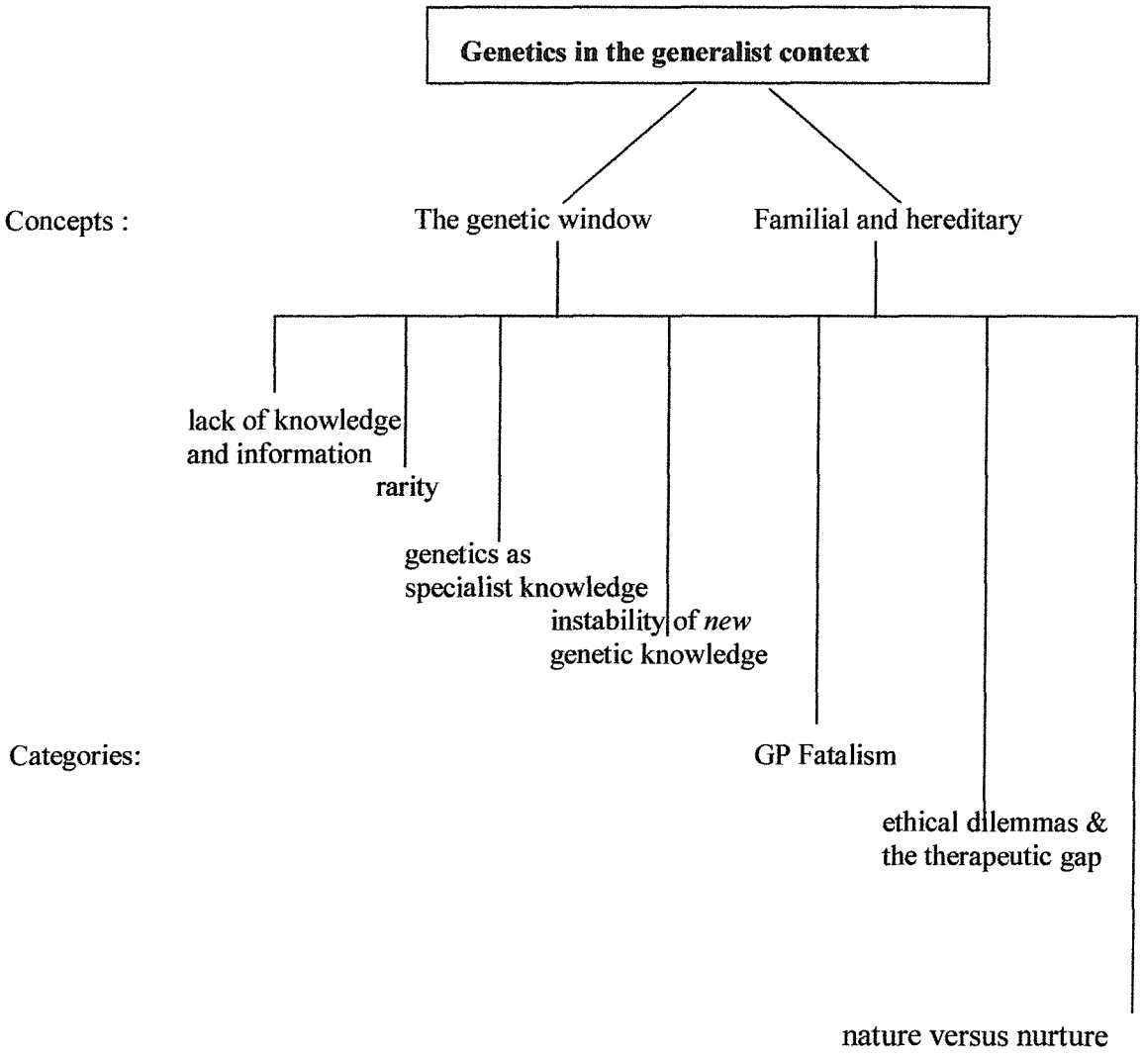
In practice these steps are not linear but iterative—sampling, data collection and analysis occurs simultaneously and so may all steps in analysis



5.4 Data and its interpretation

Below I have presented a schema for each theme showing how they are linked to the concepts and categories from which they were developed. Each schema is followed by an explanation of the categories and concepts supported by extracts from GP interviews.

Figure 5.2 Theme one genetics in the generalist context



5.4.1 Category 1: Lack of knowledge and information about genetics

"I qualified in 1959, my knowledge about genetics is rusty; we hardly got any training in medical school about genetics and I don't think I ever saw a geneticist until I went on this course." (GP 23, purposive sample).

"I qualified in 1985 and we got a few lectures on genetics but they didn't go much beyond Mendel." (GP 4, purposive sample).

"I haven't any knowledge about any new advances in genetics to do with cancer." (GP 30, an *average* GP, theoretical sample).

"When I was a medical school and when I trained in general practice the diseases were caused by chromosomes and now its genes and every bit of genetics seems new." (GP 12, purposive sample).

All respondents were consistent in reporting the lack of education, information and training they had received during undergraduates and postgraduate years that was irrespective of their year qualification. This may have enhanced GPs' perceptions of the *newness* of genetic medicine.

5.4.2 Category 2: Rarity: *"Genetic diseases are actually few and far between from our perspective."*

Overall, GPs did not consider their lack of training in, or information on genes and or their role in diseases aetiology as problematic because they rarely encountered genetic conditions in day-to-day general practice.

"These diseases are all, even cystic fibrosis, very rare, because genetic diseases are actually far and few between from our perspective."(GP 3, Purposive sample)

In this context genetic advances were seen as having little relevance for practice;

"Genetic conditions are not our bread and butter, the new genetics has little impact on my day-to-day clinical work." (GP 16, purposive sample).

"It's very difficult to say but I don't deal with genetic enquiries very often. It's not a very big part of my work, I'd expect to get asked questions in the antenatal clinic or during child health surveillance clinics but even there it's rare" (GP 7, purposive sample).

Another GP described the issue of identifying women at risk of genetic susceptibility to breast cancer in terms of the likely size of the problem for his personal list-(average list size is 1 850 patients).

"From what I can remember from the course for my own practice the number of women who carry the breast cancer gene are going to be far and few between. So I can't justify devoting a lot of time and effort on devising a system for identifying a

small number of women in the practice who might carry a gene for breast cancer. There are too many other pressing things for me to do for my patients." (GP 3, purposive sample).

A GP from the theoretical sample echoed this view:

"Genetic conditions are rare for us, the new genetics will have little impact on our patients and the kind of clinical work we do." (GP 36, *average* GP, theoretical sample).

An exception to the idea that genetic conditions were rare in general practice came from a GP whose patients' rate of consanguineous marriages were above the population average. His clinical duties included providing pre-conceptual advice to his patients for thalassaemia, sickle cell anaemia and managing other rarer genetic conditions. However, he did not think it was the GPs' role to calculate patients' genetic risks but he considered his role to be support patient decision-making, and managing the social implications of the genetic diagnosis. Often, he saw patients after their visit to the consultant clinic to re-explain their genetic risk in terms that they found more comprehensible.

5.4.3 Category 3:genetics as specialist knowledge

GPs agreed in principle that genetic advances and its clinical implementation were specialised knowledge that should be interpreted and applied by consultant geneticists.

"The new genetics is highly technical and often described in obscure language which makes it difficult for people outside that arena, like GPs, to follow the developments fully." (GP2, purposive sample).

Another GP who was interviewed as part of the theoretical sample because she held dual post-graduate qualifications in general practice and obstetrics (membership of the Royal College of General Practitioners and the Royal College of Obstetrics and Gynaecology), and had a special interest in genetics spoke about her clinical experience and knowledge as follows :

"I am not two faced about it –I am trained as a GP and an obstetrician so I have worked with couples seeking advice about pre-conceptual risks of any child inheriting disorders in the family. Enquiries were mainly about common single gene disorders, you know cystic fibrosis, occasionally muscular dystrophy and some chromosomal translocations—that's a different sort of genetics to deal with. What's being found for breast cancer where the gene acts as if it were an autosomal dominant condition but it isn't—I mean that is quite complicated for me to do as a GP without specialist input—so the genetic susceptibility to breast cancer can only be dealt with effectively by geneticists" (GP 40, GP who had completed advanced training in obstetrics and worked two sessions per week in an antenatal clinic, theoretical sample).

Another GP described the differences between her and consultant geneticists to support her perceptions of genetic medicine as specialist practice

"The specialists will be better able to deal with genetic enquires than me. They've got many expertises-first and foremost they have counselling expertise their knowledge about genes and risks." (GP 9, purposive sample).

Except for one GP, all others in the purposive and theoretical samples thought calculating a individual's genetic susceptibility to a common condition such as breast cancer, and then interpreting this risk for other family members (who may be first degree or second degree relatives) required specific specialist genetic skills. GPs who attended the *Genetics for GPs Course* were more explicit about why genetic medicine was a specialists domain. They pointed out genetic conditions were rarely managed in day-to-day general practice and so GPs would not be able to develop and sustain the degree of skill required for drawing pedigree charts and generating Mendelian fractions on which genetic risk assessment depended. Thus it was important people received risk assessments from experts who GPs thought would keep up-date with developments.

The exception was a GP who was interviewed as part of the theoretical sample because of her close professional involvement in community screening for cystic fibrosis. This GP believed that genetic risk assessment could be successfully incorporated in to routine general practice with support from a local geneticists or genetic nurse specialists. Her views were based on managing an established single gene disorder in which (one mutation accounted for 72% of cases) information on risks, screening procedures, social and ethical dilemmas had been accumulated and rehearsed over the

past few decades. This was not the case for common cancers.

5.4.4 Category 4: the instability and uncertainty of *new* genetic knowledge

GP: "These advances being made are too close to research."

SK: Can you explain what you mean?

GP: "I mean that its too new we can't be sure of it yet--we can't be sure what we're being told won't be very different tomorrow. No body knows why some women with a strong family history of breast cancer and the gene for breast cancer won't go on to develop breast cancer. We all know too well from experience that evidence we base our healthy promotion advice on this year may be challenged the next--so we end up back-tracking. Look what happened to the debate on the use of salt for hypertensive patients or for that matter the cholesterol issue.

SK: So how does what you're saying impact on identifying women with a family history of breast cancer and assessing whether they might have genetic susceptibility.

GP: Well, what I'm saying is ... if we [GPs] *are supposed to find women with strong family histories of breast cancer and tell them they're at greater risk of breast cancer than other women based on what we know now then we might be telling some women the wrong thing. I can't remember the figures but not all women with a strong family history, or with the gene will develop breast cancer—so why start worrying these women. We know family history of any cancer means relatives are more likely to develop cancer but we don't know for certain that they will, and from what I've read not every one with the breast cancer gene will develop breast cancer."* (GP 38, GP and epidemiologist, theoretical sample)

"The thing about breast cancer is that its still very uncertain what a family history means more than we already know which is people with cancer in the family are more likely to get cancer. Say I see a patient with a strong family history of breast cancer I can refer her to the geneticist who might be able to offer a genetic test if there is tissue from her relatives. And then if she has the test and its negative what does that mean--- not much she can still get breast cancer, and she can still have genes for breast cancer that we know nothing about. So it not necessarily going to change anything for the woman." (GP 3, purposive sample)

One GP had attended the genetics course because she worked as an assistant in obstetrics and wanted to learn more about single gene disorders e.g. cystic fibrosis and sickle cell anaemia. She highlighted concerns about "new" knowledge which were that it was untried and untested.

"I see anything from cystic fibrosis to Downs syndrome where I am on easier ground because the problems of communicating about it and screening for it have been so widely discussed, and you can pick that up and are easily available. You know what you're reading has been tried and tested---that level of information and discussion isn't there for conditions they spoke of on the course-- like breast cancer. I don't always remember the risks for straightforward chromosomes disorders but at least I can look them up quickly and find out the screening procedures that's just not there for the other conditions [common cancers]." (GP 9, purposive sample)

These quotes illustrate the uncertainty general practitioners felt about the reliability and stability of emerging knowledge on genetic susceptibility to breast cancer. They thought the *instability* of new genetic knowledge was a sound reason for not changing their current practice. This was based on GPs' experiences of how knowledge in other health contexts e.g. salt and hypertension, and fat in-take and cardio-vascular disease had changed over time with consequences for how their authority, credibility and professionalism was perceived by patients.

5.4.5 Category 5:GP fatalism

Whilst some GPs were reluctant to identify women with genetic susceptibility to breast cancer because they did not believe in the certainty of the evidence, others were reluctant to identify women because of more fatalistic thinking about inherited breast cancer.

"What can women do if the have the gene for breast cancer—nothing so there is no point in giving people a death sentence. So I wouldn't alert women to their risk." (GP 5, purposive sample)

"I don't think I will be identifying anyone with a family history of breast cancer –its futile. You'll just create a very anxious group of people who will live in fear about getting cancer and for whom we can't alter the risk" (GP 18, purposive sample).

For these GPs, their thinking appeared to be grounded in ideas of genes as deterministic—that genetic susceptibility was difficult to influence by changing life-

style or environment-- genetic endowment was seen as immutable.

SK: What are your thoughts about gene expression and life-style

"I suppose we might be able to influence gene expression...that is where things like smoking where you need two effects to get a disease. You might need two accidents inheriting a gene to make you susceptible and the other might be environmental that triggers it off, but I do not understand with each illness how big each of those factors are, so I would not express it as changing your genes. I would say there is the genetic bit that we cannot help but there is the other bits that somehow work with that, that we can alter, so I would not be scientific about it I might actually try... and that is how I would try and talk to the patient about this" (GP 26, purposive sample).

Another GP spoke of making people comfortable with the genes they lived with by utilising quite radical procedures such as mastectomy.

"I think there are risk factors in the environment that you can reduce for that patient. They can live with the gene more comfortably. I'm not sure how that applies to breast cancer but I suppose you could even advocate in certain situations, if it was, I mean, enormously high risk you could contemplate mastectomy." (GP 40, professor of epidemiology in general practice, theoretical sample).

Other GPs expressed their fatalism by comparing genetic susceptibility to breast cancer to people infected with HIV who had developed auto-immune deficiency syndrome (AIDS).

"It's a bit like HIV at the moment—because once you've had the gene test and its positive you'll be waiting for the condition to develop" (GP 7, purposive sample).

GPs also speculated on how they may record future gene tests results for common conditions such as cancer. Again some of the GPs who attended the genetics for GPs course compared recording patients' gene test results to recording patients' HIV status, and to dilemmas about recording patient requests for HIV counselling. As with HIV some GPs reported they would not record gene tests in notes with the patients request and would refer requests for results of gene tests from third parties such as insurance companies directly to patients themselves.

"All this reminds me of aids and how we had to be careful of what we put in the notes—I've only seen three people myself but I did ask them if it would be ok to write it in the notes." (GP 17, purposive sample).

Other GPs from the purposive sample did not think recording peoples' gene tests was a problem because a strong family history of breast cancer would be seen as conclusive for positive carrier status. One GP from the theoretical sample, with personal experience of a genetic condition, was concerned that GPs be more careful about recording family history in medical notes. She thought family history would eventually predispose insurers, mortgage companies and employers negatively to people with strong histories of cancer.

5.4.6 Category 6:Ethical dilemmas and the therapeutic gap: *"I don't raise the issue of genetic risk unless I think there is something positive that can be done."*

Surprisingly, given the prominence of the new genetics, all respondents thought that genetic advances would have little impact on their management of common conditions. However, they arrived at this position for different reasons. Informed GPs who were aware of the on-going genetic re-definition of common diseases (e.g. BRCA1 and 2 conferring susceptibility to breast and ovarian cancer), were reluctant to alert patients to genetic risk in the absence of effective screening technologies and therapies to reduce risk or prevent disease-in other words they identified a therapeutic gap which in their minds operated as an ethical dilemma.

"The problem with these diseases (breast and ovarian cancer)...what can you do about it if you've got the gene. Is there any point in giving young patients a death sentence. You must be able to alter the natural history of the disease and if at the moment we can't do that effectively then I can't see myself raising the issue." (GP 26, purposive sample).

Here, this GP is voicing concern about sharing information with the patients and so allowing the patient to make a choice. The assumptions being made by the doctor are paternalistic. Paradoxically, this doctor framed her reticence to discuss genetic risk as 'ethical,' a use of that term which appears to refer to making decisions on the patient's behalf.

Another reason GPs gave for not raising the issue of genetic risk, was the anxiety they would create for patients who might then seek additional advice from practice nurses and practitioners.

" I don't raise the issue of genetic risk for common conditions unless there is something positive that can be done, which there isn't and anyhow even if I did it would only make them and their family anxious and so possibly more work to do." (GP 34, GP with special interest in genetics, theoretical sample).

"The breast cancer things is different because not all people with a strong family history of breast cancer will be able to have a test so to talk about it will be just like opening-up a can of worms for all of us. which is what this is all about really." (GP 11, purposive sample).

5.4.7 Category 7: nature and nurture

GPs recognised too the potential tensions patients and families face in managing behavioural/lifestyle changes in the context of deterministic genetic knowledge:

"It will be very difficult for patients to balance the finality of carrying genes for heart disease or cancer and then receive advice to give up smoking, take exercise and increase fruit and vegetable intake. How do genes and lifestyle interact, will adopting a healthy lifestyle change their genes, their genetic risk-these are the things I need to know" (GP 3, purposive sample).

5.4.8 Concept 1: appropriate generalist intervention: *"the genetic window."*

It was in the context of established genetic diseases that GPs were clearest about their role--using family histories collected in specific circumstances (pre-conceptual or ante-natal, and child health surveillance clinics). GPs who did not identify a genetic component to common cancers saw the genetic advances impacting on established genetic conditions e.g. cystic fibrosis.

"Most of these advances will be for improving our treatment of diseases like cystic fibrosis or screening for Down's syndrome." (GP 5, purposive sample).

"The times that I am most likely to collect a family history in a more comprehensive way is during antenatal clinic or if I'm lucky during pre-conceptual consultations" (GP 9, purposive sample).

Common diseases were described as multifactorial in cause, but GPs spoke only in terms of lifestyle, nutrition, and environmental toxins and not genes.

"For cancers I usually try and make people aware of what lifestyle changes they can make. I don't so much talk figures but say cut down on smoking, eat more fruit and vegetables, eat less saturated fat and take exercise-things like that, healthy advice." (GPs 11, purposive sample).

The existence of multiple cases in a family were interpreted as exposure to common carcinogen(s) rather than shared genes. These GPs collected family history to gain insight into the possible psychological and social impact the disease had on family members.

" For women with a strong family of breast cancer I would ask how old their mother was when she got the breast cancer, I would talk to them about the fact it was common and I would want to know this in order to understand if they knew what the cancer could do to them, it was not to find out if they were at risk of cancer." (GP 17, purposive sample).

5.4.9 Concept 2: the familial: hereditary distinction.

This analysis led me to explore how GPs differentiated between the terms familial and hereditary disease in the context of common diseases. Familial was consistently used to describe conditions shared by family members who were not genetically related such as husband and wife.

"I would take familial to mean conditions that crop up in the family in relatives who aren't related, for example, I see depression, alcoholism and obesity affecting husbands and wives." (GP 41, professor of general practice, theoretical sample)

"Cultural and environmental on the one hand and genetic inherited characteristics on the other. That is how I distinguish between genetic and familial if you like, or

hereditary and familial in that I would think of hereditary as being genetic probably, and familial just being running in the family but I would need convincing that it is genetic, although I know with some people it is. I think that was cultural/environmental." (GP 37, GP principal and professor of general practice, theoretical sample).

"We record family history at some point on most days but we don't record it to be used a genetic screening tool." (GP 28, purposive sample).

" When we register patients we get them to fill out a form which is filed in their notes. The form asks about their family history; cancer, hypertension, diabetes, stroke, asthma, eczema and so on. These things run in families that's why we ask about them"
SK: Why do you think these conditions run in families?
"It's an observation which hasn't got a scientific basis at present unlike for things such as polyposis coli which is genetically inherited." (GP 33, GP teacher, theoretical sample).

"I record family history when patients first register—Is your father well? Has he got any illnesses? Is your mother well? Has she got any illnesses? Have you got any brothers and sisters and how are they? So I get the diseases written down—we like to know?
SK: Do you ever ask the age of onset of the illness in other family members?"
"No, you know in a 10 minute consultation there's no time for that—if we need that

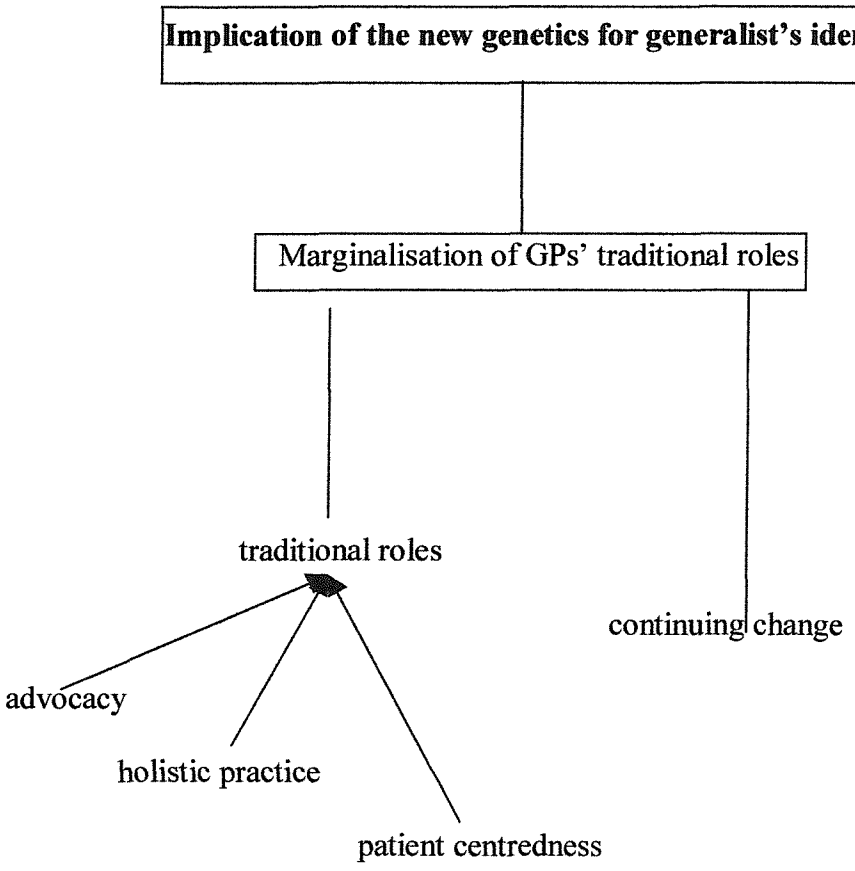
sort of information then I could arrange for it to be collected by the practice nurse during the new patient check." (GP 19, purposive sample).

Hereditary on the other hand was consistently reported as implying shared genes. This distinction was not made by the consultant cancer geneticist or the professor of genetics, interviewed as part of the theoretical sample, who used the terms synonymously to signify shared genes. However, informed GPs used 'hereditary breast cancer' to distinguish women where genes exert a powerful effect. 'Familial breast cancer,' on the other hand was taken to mean exposure to shared environmental carcinogen(s).

In making this distinction, GPs begin to address the issue of recognising common diseases as genetic or having a genetic component. This is crucial if they are to use family history to assess genetic risk. The director of the genetics interest group has pointed out "patients do not go to the doctor saying; 'I think I have genetic disorder,' its a matter of getting the light to come on in the GP's head saying 'I wonder if this is genetic."

5.5 Theme two: Implications of the new genetics for generalist identity

Figure 5.3 Theme two: Implications of the new genetics for generalist identity, *'even more change.'*



5.5.1 Category 1: Continuing change

"Genetics is an expanding field, its not that long ago when it was just a lab subject it's only now that it might be useful to us. As far as general practice is concerned it's going to mean even more change" (GP 9, purposive sample).

All respondents perceived the new genetics as another in a series of changes to be

imposed on general practice. This perception was reinforced by policy makers and experts emphasising the new genetics as a series of additional tasks requiring new knowledge and skills, rather than an extension of current practice. Overall, GPs in the purposive sample tended to view implementing genetic advances in a negative light but GPs in the theoretical sample who had a special interest in genetics were cautiously enthusiastic. They looked on implementing genetic advances as an opportunity to escape from what they described as the mundane and repetitive aspects clinical general practice. The average GPs who formed part of the theoretical sample echoed those in the purposive sample. However, only one GP, interviewed in the theoretical sample because of her experience in implementing cystic fibrosis screening in the community advocated taking on the role of identifying people at genetic risk of developing common cancers irrespective of ethical issues surrounding the absence of effective screening and therapeutic interventions.

5.5.2 Category 2 traditional roles

"The traditional role of GPs is doing what GPs do and that is defined in our service agreement. GPs haven't traditionally looked after people with chronic renal failure, traditionally done warfarin clinics and certainly haven't done genetic counselling" (GP 18, purposive sample).

"I feel my contribution is knowing which key opens the door, which direction to send people for specialist advice about their family history. I don't regard myself as having much expertise despite attending the course. I wouldn't regard that as having made me

an expert in the field. What that course did show me was how complicated the new advances about breast cancer are and that it would be incredibly complicated to give advice on that. It has to be something you do day in day out" (GP 2, purposive sample).

The average GPs in the theoretical sample concurred with the views expressed by the two GPs above. Again GPs with an interest in genetics saw this as an opportunity to make GPs' roles more specialised and delegate tasks such as providing care for people with acute minor illnesses to practice nurses. The GP with experience of cystic fibrosis screening in the community, the professor of genetics and the consultant geneticist were the most enthusiastic about GPs taking on new roles.

5.5.3 Concept: marginalisation of generalism

GPs from both samples highlighted how the new genetics tied in to the on-going debate about the nature of generalist practice and its core values which is fuelled by continuing contractual changes. These GPs resisted taking on genetic risk calculation for common diseases because it challenged their perception of themselves as autonomous providers of holistic and generalist medical care.

'Because genetics has long term implications for the family primary care is inevitably called in. For example, my role in genetic counselling would be supportive dealing with the implications for the family and the children. I don't want to calculate genetic risk'. (GP 7, purposive sample)

GPs were concerned that additional specialist skills could threaten what they perceived to be their traditional roles and core skills.

'GPs are sick to death of being asked to do traditional secondary care as primary care...where do we get the time to see our normal patients and do what GPs traditionally do.' (GP 4, purposive sample).

Another GP said; *'My strength lies in knowing who to refer to. I am not an expert on genetic counselling or calculating risks but I can help in explaining things to patients once they've seen the geneticists, and I can help them through the social and psychological effects.'* (GP 17, purposive sample).

Whilst some of the GPs expressed resistance to the idea of identifying people with genetic susceptibility to common conditions they also showed in their responses they were aware of some important issues raised by identifying people, providing people with risks, and recording these risks in patients' notes. GPs spoke mainly in terms of the clinical, and ethical dilemmas they may face but were less explicit about the psychological and sociological implications of identifying patients' genetic susceptibility to common conditions. This was true for most GPs in the purposive and theoretical samples—however GPs did think they would be best suited to managing these dilemmas.

5.6 Summary

The qualitative analysis highlighted the differences between what experts thought GPs role should be, and general practitioners' ideas of their role. General practitioners emphasised the need to build on their current practice, whereas policy-makers and experts focused on transforming, or geneticizing general practice by advocating new specialised roles. Two themes were identified, *genetics in the generalist context* and *the implication of the new genetics for the generalist identity.* The first theme included: appropriate generalist intervention, instability of genetic knowledge, ethical dilemmas and the "therapeutic gap," and family heredity distinction The second theme: implications for the generalist identity included the marginalisation of generalism. The two themes support that the genetic advances requiring implementation in general practice should be integrated within the existing generalist framework.

In the next chapter I discuss how a hypothesis generated for the category *family history and heredity* was used to generate a questionnaire for application to a random sample of GPs.

Chapter Six

Linking Data: the development of the GP survey, the results and their analysis

6.1 Introduction

This chapter describes the development and execution of the general practitioner survey, and begins by clarifying the rationale, aims, and objectives of the survey and continues with a description of how the self-administered questionnaire was constructed. Specific attention is paid to how the GP questionnaire was developed to collect data on two hypotheses generated from the qualitative study—thus making transparent how the qualitative and quantitative studies were linked. The later sections present the survey method followed by a descriptive account of the survey results and finally a factor analysis of the results.

6.2 Rationale for survey research

Survey research has been defined as, "*a set of scientific procedures for collecting information and making quantitative inferences about populations*" (Oppenheim 1995). This involves systematic gathering of information from a sample of respondents using a survey questionnaire, which is, *'an instrument consisting of a series of questions and /or attitude-opinion statements designed to elicit responses, which can be converted into measures of the variable under investigation'* (Franklin and Osborne 1971). The information collected by a questionnaire can be converted into meaningful numbers by classifying, counting and scoring, followed by summarising and estimating opinions, behaviours and attitudes in the sample or the

population from which the sample was drawn. Broadly speaking there are two main types of survey: 1) descriptive, enumerative or census type which tend to be fact finding and 2) the analytical relational type. This survey falls mainly into the first category, as the primary goal was to enhance the understanding gained by the qualitative study and not to generate complicated statistics of great analytical precision.

6.2.1 Self-administered questionnaires

Self-administered questionnaires are instruments used to collect information from people who complete the questionnaire themselves. An important advantage of self-administered questionnaires over supervised questionnaires is their delivery by post. This confers several other advantages, briefly these are: lower administrative costs, wider geographical coverage, easier access to larger sample sizes, easier implementation, and more control over timing (Bourque and Fielder, 1995). For these reasons, the GP survey was designed as a postal self-administered questionnaire study. The disadvantages of self-administered questionnaires include less control over the response rate, limitations on the number of questions and limitations on the complexity of questions.

6.2.2 Rationale for using qualitative data to construct questionnaire items

When qualitative research is undertaken as a precursor to quantitative research it often acts *"as source of hunches or hypotheses for the quantitative research"* (Bryman 1988). In the context of designing the GP questionnaire, insights gained from the qualitative study acted to minimise ambiguities in the language and

categories used in the questionnaire. For example, the qualitative study showed that many GPs classified genetic conditions, such as cystic fibrosis, as 'hereditary,' and common late-onset conditions such as breast cancer as 'familial.' For these GPs, *familial* and *hereditary* had distinct meanings that were not fixed but were contingent on the clinical context (see chapter five). In contrast, clinical geneticists used both terms synonymously to indicate a genetic foundation to any condition. This observation offered an insight on how to avoid GPs' misunderstanding the categories deployed in questionnaire items--the boxed text below helps to demonstrate this point.

When GPs discussed family history in the context of cancer they tended to view the significance of *family history* for the information it provided about an individual's or family's life-style, social and psychological contexts. This was in contrast to genetic conditions, such as cystic fibrosis, where family history was primarily viewed as a source of genetic information. Thus, by providing an explicit clinical context, GPs' opinions of the clinical utility of collecting the different aspects/categories (see figure 6.2) of family history could be gauged.

Conducting a qualitative study before constructing a questionnaire offered other pragmatic and theoretical advantages: the identification of issues by GPs which they considered pertinent to their practice; the identification of issues and concepts not foreshadowed in existing literature (see 6.7.1); the provision of contextual and theoretical information which could facilitate the interpretation of the GP survey (see 6.7.3); and a source of terminology used by GPs with the potential to increase the comprehensibility of the survey items. These advantages, in turn, helped to

maximise the face, content, and construct validity of the questionnaire items (Bryman 1988).

6.3 Aim of the GP postal questionnaire survey

The overall aim of the questionnaire study was to survey a representative sample of GP principals on the clinical utility of exploring different dimensions of a family history of cancer. This aim reflects two of the main hypotheses derived from the qualitative study (see chapter five):

1) When GPs collect a family history of cancer their primary aim is to assess the social and psychological impact of the cancer on the patient and their family.

2) GPs regard a family history of cancer as indicating exposure to shared environmental and dietary carcinogens rather than shared genes.

6.3.1 Objectives of the postal questionnaire

Based on these hypotheses several specific objectives were defined:

1. To collect data on GPs' personal and practice attributes: age, sex, professional qualifications, year of qualification, type and site of practice.
2. To measure the number of GPs who collected family history to foreshadow social problems.
3. To measure GPs' opinions of a family history of cancer as signifying the cancer's aetiology.
4. To measure the extent to which GPs regarded family history as an indicator of genetic susceptibility.
5. To measure GPs' belief that a family history of cancer provides information about lifestyle risks for developing cancer.

6. To measure GPs' opinions of family history as tool for assessing genetic risk in other family members.

6.4 Questionnaire construction

Established principles of questionnaire design guided the construction of the questionnaire (Oppenheim 1995, Dillman 1978). At base, these principles are simplicity, intelligibility, clarity, accessibility and brevity. The items were worded to reflect these qualities which were made more achievable because of the preceding qualitative study. The content and the scope of the questionnaire were determined by the need to secure a good response rate and by the objectives of the qualitative study. The different stages involved in producing the questionnaire are described below:

Stage 1) Delimiting the scope of the questionnaire by selecting specific themes from the qualitative data for generating questionnaire items and establishing construct validity.

Stage 2) Deconstructing the core theme to develop questionnaire items.

Stage 3) Testing the validity and reliability of the questionnaire.

Stage 4) Piloting a prototype questionnaire—establishing face validity and content validity.

Stage 5) The final version of the questionnaire.

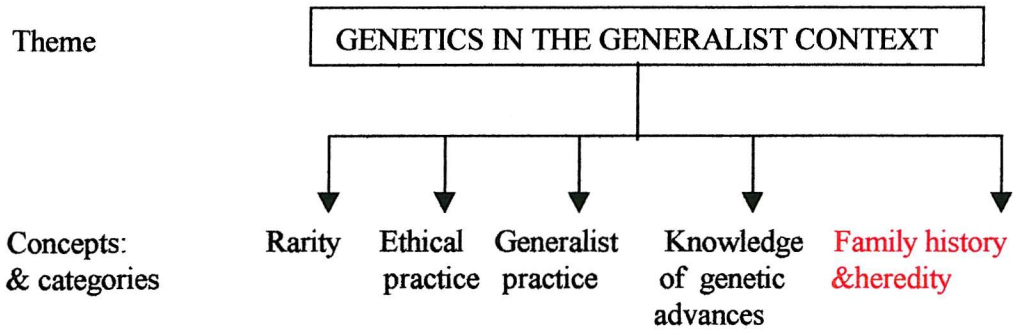
6.4.1 Stage 1: delimiting the scope of the questionnaire

Securing a good response rate was an important factor in determining the scope of the questionnaire. In general, in the context of self-administered postal surveys, an

inverse relationship exists between the response rate and the questionnaire length. A lengthier questionnaire requires more time to complete, and is thus less likely to be completed. For this reason, a brief questionnaire was developed. Consequently, it could reflect only a fraction of the qualitative analysis.

One of the core themes, *genetics in the generalist context* (see figure 6.1) acted as the foundation from which questionnaire items were constructed. This theme was selected because it encapsulated the most significant concepts and categories derived from some of the most robustly tested data collected from the purposive sample, and then explored further by careful theoretical sampling.

Figure 6.1. Deconstructing the core theme



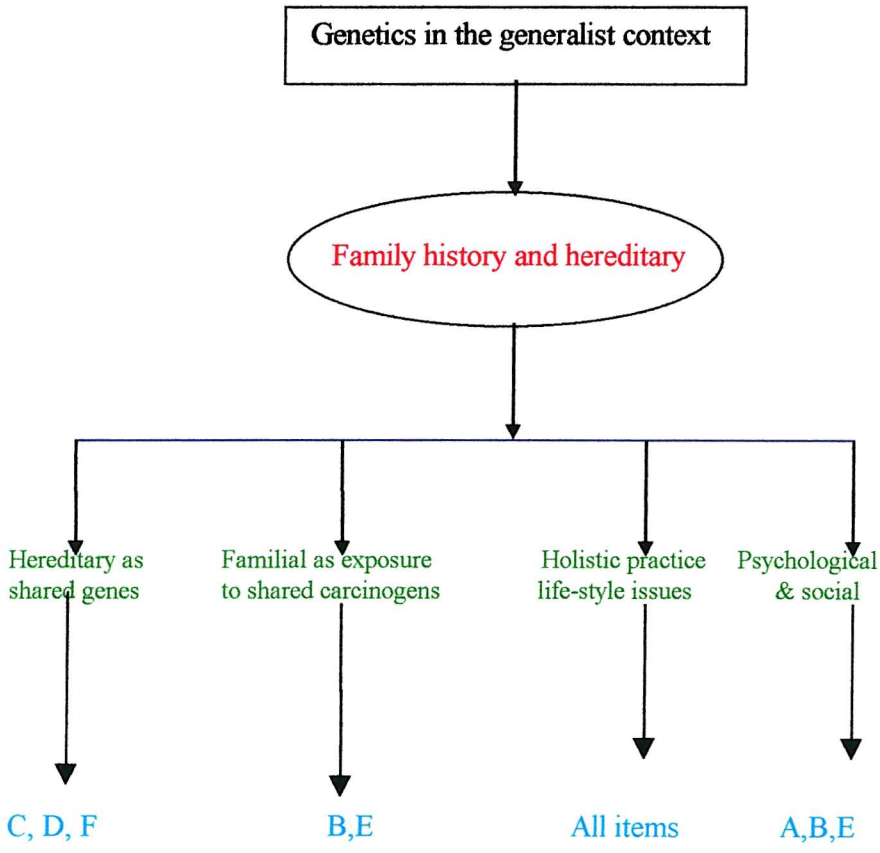
To operationalise the theme into questionnaire items it was first reduced to its constituent concepts, categories, and sub-categories (see figure 6.1 and 6.2). The theme consisted of a number of concepts (shown above) which in turn consisted of a number of categories derived from the interview data. Thus, the questionnaire items could have been developed to include any one or all of these. However, only one concept, *family history,* formed the basis of the questionnaire. This was done for a number of reasons. Firstly, the qualitative study had identified variation in how family history was understood and deployed by GPs in different clinical contexts as an important issue. Secondly, the appropriate collection of family history is identified as an essential skill to allow GPs to assess genetic risk and manage patients and families appropriately (Austoker 1994). Thirdly, the issues of how the concepts 'familial' and 'hereditary' operated in general practice were not foreshadowed by existing literature. Finally, the aim of securing a good response rate necessitated keeping the questionnaire as brief as possible.

6.4.2 Stage 2, deconstructing the concept of *family history*

Figure 6.2: operationalising *family history* as questionnaire items

I have used a colour code to signify different levels of data

Theme	Boxed black text
Concept	Red
Categories	Green
Specific questionnaire items	Turquoise



Letters refer to questionnaire items see appendix 2

An explanation of this theme and its construction is provided in chapter five.

When this questionnaire was designed, there were no published surveys of British GPs' knowledge about advances in cancer genetics nor data on their opinions of the implications advances would have on their clinical practice. Neither was there

recently published evidence on GPs' use of family history to assess genetic susceptibility when a patient was diagnosed with cancer. However, the findings of the qualitative study showed that some GPs did collect family history when a patient was diagnosed with cancer, but their primary aim for doing so was to assess the patient's and the family's ability to cope with the psychological and social consequences of the illness. On the whole, GPs were uncertain of the family history details they needed to collect in order to assess the genetic risk to other family members. The significance of this finding has been discussed in chapter five.

6.4.3 Stage 3: reliability and validity in survey research

The development of reliable and valid measurement instruments is an important objective in research because it is generally considered essential to be able to judge the extent to which results can be attributed to random or systematic error. Reliability refers to the consistency or stability of a measure or test from one use to the next and, tests of validity concern the accuracy of measurement (Litwin 1995). Researchers are encouraged to draw upon previously validated instruments whenever possible; however, in the context of measuring GPs' opinions on implementing genetic advances there were no pre-existing validated instruments at the time this survey was designed. Even if there had been an instrument available from another speciality, its suitability for application would have needed review because of the importance attached to context in primary care. This is such that the qualitative perspective--that what matters is authenticity rather than reliability--seems more convincing than the quantitative one, that the conditions for an enquiry can be reproduced, as for example in pre- and post- test reliability testing.

6.4.4 Stage 4: Piloting a prototype questionnaire to assess face, content and construct validity

Piloting questionnaires and assessing their face, content and construct validity of questionnaires are essential steps in the process of refining a questionnaire (Dillman 1978).

Face validity—*whether 'on the face of it', the questions are measuring what they are supposed to measure.* I assessed face validity informally by asking five service GPs and five academic GPs to consider whether the questions appeared relevant. All responded positively.

Content validity—*whether the choice of items and the relative importance given to each is appropriate in the eyes of those who have some knowledge of the topic area.* I assessed content validity by posting a draft questionnaire to six "informed" service GPs. Four GPs were selected from the purposive sample and two were identified from *The Practitioner*. This is a monthly journal where general practitioners discusses the clinical management of challenging medical conditions from a perspective deemed useful to other general practitioners. In July 1996 and February 1997 *The Practitioner* published articles which discussed the genetic basis of breast cancer and GPs' management of a family's history of breast cancer (Walker, Berry and Rose 1996, Higson, Cembrowicz and Baum 1997). Additionally, three senior GP academics experienced in questionnaire design were asked for their opinions. Between them, these GPs practised in urban, semi-rural and rural practices in Somerset, Hampshire, and Wiltshire.

I sent the questionnaire to this group of nine GPs with an explanatory letter inviting them to make comments on whether they thought questionnaire covered everything it should and to point out extraneous material. All replied with comments suggesting changes to the wording or the focus of questions in order to maximise clarity, the addition of tick boxes to reduce completion time by avoiding unnecessary writing and, queries about the use of technical language. For example, one of the questionnaire items focused on GPs' collection of *genetic data* in different clinical contexts. All of the GPs found the term *genetic data* confusing and did not equate it with family history. All comments were reviewed carefully and changes were made to the questionnaire.

Construct validity—*whether the results obtained using the questionnaire confirm expected statistical relationships, the expectations being derived from underlying theory.* For example, when constructing questionnaires researchers make assumptions about the nature of categories as well as how they will relate to each other. It is precisely here that a qualitative study can provide a theoretical basis (acting as the *underlying theory*) from which such assumptions can be derived. Thus, in light of the findings of the qualitative study, the GP survey would be expected to show the following patterns. GPs who collect family history to assess the role of life-style and environmental factors on cancer susceptibility are more likely to use family history to assess the psychological and social impact of cancer on the family and the individual. Conversely, GPs who recognised family history as a source of genetic information were less likely to use it to assess life-style and environmental factors or assess the psychological and social impact of cancer on the individual or their family.

6.4.5 Stage 5: Developing the final version of the questionnaire

Final changes were made to the questionnaire to reflect all the feed-back received during the developmental phase. A copy of the final questionnaire appears in appendix 2.

6.5 The Survey Method--calculating the sample size

In general the larger the sample size the smaller is the random sampling error. Thus to detect small differences requires large sample sizes. However, pragmatic considerations such as cost and feasibility often dominate and these were the limiting factors in my work.

6.5.1 Subjects

How well a sample represents a population depends upon the sample frame, the sample size, and the specific design of the selection procedures (Fowler 1998). In this study, the sampling frame consisted of GP principals working in the geographical area served by the Wessex Regional Health Authority[1] (see footnote on page 160). It was assumed that GPs were more likely to respond to a postal survey originating in their own region. The most up-to-date list of GP principals on CD-ROM form was obtained from the Wessex Regional Health Authority. This acted as the sample frame from which a simple random sample of 200 general practitioner principals was generated by computer.

There are limitations to the lists kept by the regional health authorities relating to list completeness e.g. the inclusion of new GP principals and the deletion of those no

longer practising. Thus, it was likely that newly qualified GPs, who would be more familiar with recent developments in genetic medicine, could have been under-represented in the sample.

6.5.2 Minimising non-response

As part of a strategy to minimise non-response, a letter of introduction (see appendix 3) accompanied the questionnaires and addressed specific processes which individuals experience when they receive a postal questionnaire. These are recognition (what is this package?), assimilation (what am I being asked to do?), evaluation (what are the pros and cons of co-operation?) and decision (whether to fill it in?) (Bergman, Hanve and Rapp 1978). Thus, the letter explained why the survey was worthwhile, that filling in the questionnaire was not excessively time consuming, that the survey was confidential, and that GPs' responses were important. The letter was printed on departmental headed paper carrying the university logo. My contact details (e-mail and telephone number) were provided at the end of the letter should GPs have any queries about the survey. A copy of the letter appears in appendix c.

The questionnaire, covering letter and an addressed free-return envelope were posted first class to all 200 GPs. A reminder was sent two weeks afterwards and a final reminder--containing the covering letter, the questionnaire, and a free-post return envelope was sent out six weeks after the first posting. Following this practice has been shown to improve the final response rate (Mangione 1995).

[1] At the time the sample was generated the Wessex Regional Health Authority was still operative.

6.5.3 Monitoring the progress of the questionnaire study

Survey operations need to be monitored and managed as part of their quality control (Dengler 1996). Because this survey involved a relatively small number of GPs, I was able to do this myself with the aid of a computer flow chart that listed all the GPs in the sample and recorded the date the questionnaires were posted to them. To preserve confidentiality the list consisted of numbers between one and 200. Each number represented one GP and corresponded to the number printed on the questionnaire sent to that GP. This allowed me to identify those who had responded and those who required reminders.

Data from the returned questionnaires was coded and entered into SPSS version 9.0 for analysis by myself. Data was entered twice to minimise data entry errors.

6.6 The results of the GP survey

6.6.1 Response rate

Completed questionnaires were returned by 135 of the 200 GPs in the random sample. Additionally, ten questionnaires were returned uncompleted with a note from the practice manager explaining the non-response. Reasons included: annual leave, illness, maternity leave, change of practice, sabbatical, death and retirement. The overall response rate was thus 135/190=71%, which is generally considered a good and reliable response (RCGP1994). Of the GPs who responded: 79 were male (see table 7.1), the majority,

82% (n=111) had graduated between 1971and 1990 (see table 7.2) and 59% (n=80) possessed MRCGP. The respondents' personal list sizes varied between 800 and 2 200 and 59% (n=80) worked in GP training practices. Thirty-five percent (n=67) of GPs described their practices as urban, 19% as (n=37) semi-rural and 16% (n=31) as rural.

Table 6.1 Respondents: percentage of males and females.

	Frequency	Percent	Cumulative percent
Female	56	41.5	41.5
Male	79	58.5	100.0
Total	135	100.0	

There were no missing data for these categories.

Table 6.2 Year of graduation

	Year of graduation					
	1951-1960	1961-1970	1971-1980	1981-1990	1991-2000	Total
Total count	3	17	58	53	4	135
Percentage	2.2%	12.6%	42.9%	39.3%	2.9%	100.0%

There were no missing data for this category.

6.6.2 Non-respondents

A pertinent question to address was why 55 of the GPs had not responded. By examining how non-responders differed from responders, it would be possible to gauge the degree of bias in the survey. Ideally, for reasons of generalisability the differences between the two groups should be minimal; if they are not, one needs to consider how any differences influence the applicability of the findings beyond the sample studied. Harrison (1977) explored the extent to which non-response can lead to bias in a study that carried out a comparison of physicians who did or did not respond to a postal questionnaire. He found that when response rates of more than 75% are achieved for a postal questionnaire applied to a relatively large sample within a designated profession or occupational group, then the differences between responders and non-responders is minimal and "unbiasing."

This survey fell just short of 75% which made it important to compare the responders with non-responders. I wanted to know how the non-responders differed from the responders and why they had chosen not to complete the questionnaire. Because the non-responders formed a relatively small group I telephoned all 55 GPs and collected the following reasons from them:

a) Ten of the 55 non-responders thought the research area was not relevant to their day to day practice—all of these GPs had qualified between 1951 and 1971 which distinguished them from the majority of the respondents who had qualified between 1971-1990.

b) Five non-responders stated they had a personal policy not to complete research questionnaires because they received too many. One of the five felt very strongly, and said it was not see part of his "NHS duties" and was unwilling to complete questionnaires unless he was remunerated.

c) The remaining 39 GPs cited *a lack of time* associated with a range of reasons e.g. high clinical demand, increased administrative task linked to practice reorganisation, and occasionally due to staff shortage within their practices.

A question on whether GPs worked full or part time was omitted from the questionnaire because it was assumed that number of hours a GP worked was unlikely to influence their responses to questionnaire. Therefore it is unclear whether *a lack of time* was related to whether a GP was part-time than full-time. In retrospect, this information would have been useful and would have shed light on the over or under-representation of each group.

Non-responders and responders were similar for most of the categories on which information was collected and so on the whole the level of bias within the sample was probably small. Table 6.3 gives characteristics of the non-responders.

Table 6.3 Characteristics of the non-responders, n = 55

Sex		**Practice**		**Location of practice**		
Male	**Female**	**Training**	**MRCGP**	**Urban**	**Semi-rural**	**Rural**
34	11	23	22	40	9	6
61%	20%	42%	40%	72%	16%	11%

6.6.3 Descriptive analysis of the survey

The next few sections present a description of the survey findings followed by a factor analysis. The results of the survey are summarised in table 6.4 below. The categories have been collapsed so that responses falling into *strongly agree and agree* form one category, *neither agree nor disagree,* and *don't know* the second category, and *strongly disagree* and *disagree* the third category. This process allowed the data to be presented more succinctly. The table is structured so that the main question is followed by the options *a-f.* Then for each option the GPs' responses to each category are enumerated.

6.7 Summary of questionnaire results

Table 6.4 Summary of GPs' responses to the questionnaire items n=135

Question : When a patient under the age of 50 years is diagnosed with cancer, finding out about their family history of breast cancer.....

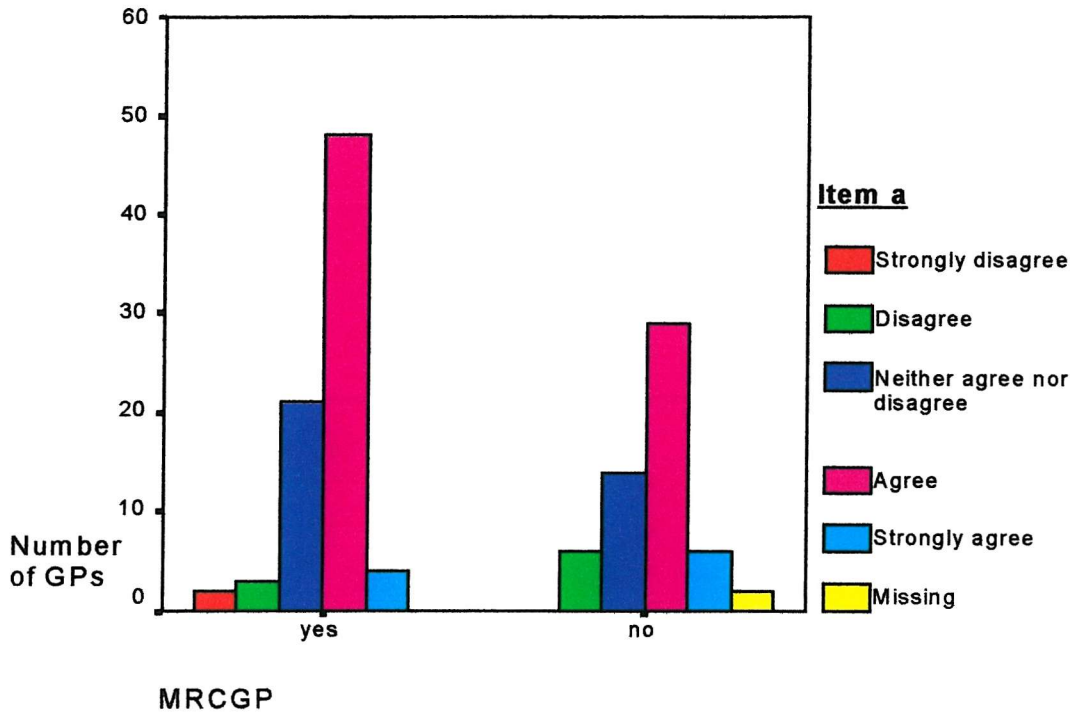
Strongly agree/ and agree	Neither agree nor disagree/ Don't know	Strongly disagree/ disagree	Missing
a) Will give me ideas about the social problems that might arise in the family			
88 (65%)	35 (26%)	11(8%)	1(0.7%)
b) Will **not** give me insight into the possible psychological problems that may arise			
12(8.8%)	139(9.6%)	**108(80%)**	2(1.4%)
c) Will provide an explanation why the patient has developed cancer			
65(48%)	55(40%)	13(9.6%)	2(1.4%)
d) Will give genetic information about why the cancer developed			
58(43%)	53(39%)	21(15.5%)	3(2.2%)
e) Will give me information about the family's lifestyle risks that may explain the occurrence of cancer			
78(58%)	38(28%)	18(13%)	1(0.7%)
f) Will allow me to assess if other family members are at risk			
114 (84%)	9(6%)	10(7%)	2(1.4%)

A brief description of the findings for each item is given below.

6.7.1 Item *a*—ideas of social problems

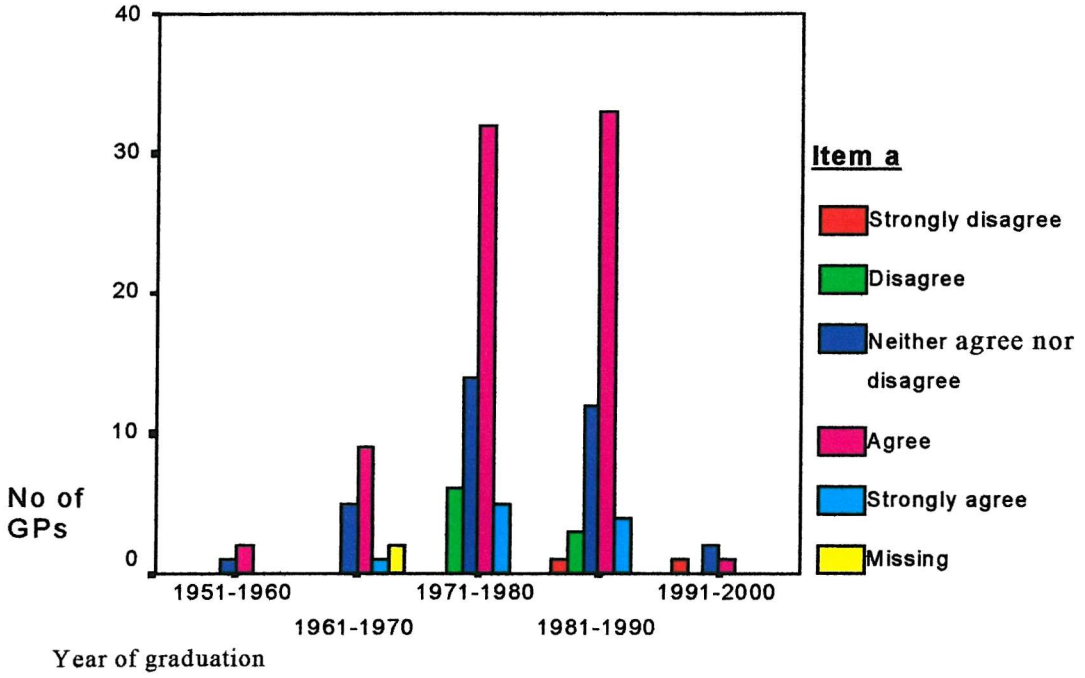
88 (65%) of the GPs *strongly agreed or agreed* that a family history of cancer would give them ideas about potential social problems. However, 35 (26%) of the GPs were uncertain and 11 (8%) *strongly disagreed and disagreed.* Closer examination of the data showed that possession of MRCGP did not change GPs' pattern of response to item *a*— this is illustrated in graph 6.1 below.

Graph 6.1 Responses to *item a* with respect to MRCGP.



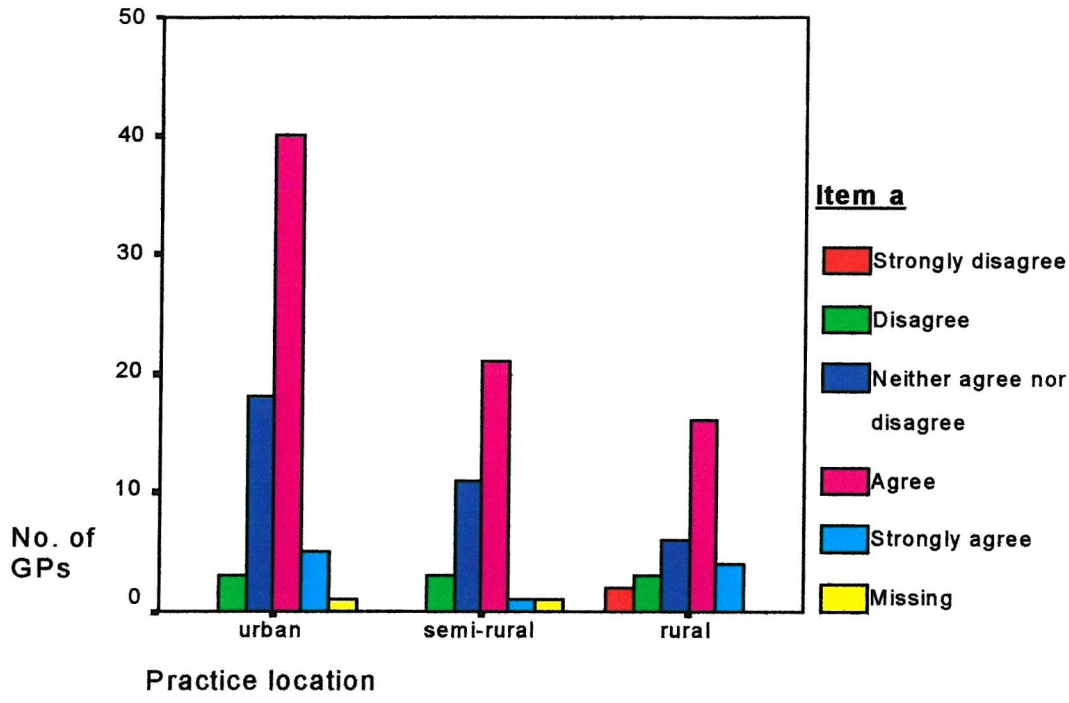
This graph suggests that GPs with and without the MRCGP qualification answered the item similarly. In both groups, the majority of GPs agreed that collecting a family history in the given context would give ideas about possible social problems. Points of difference between the two groups related to two of the GPs with MRCGP group *strongly disagreeing* with the statement and two GPs without MRCGP not responding to this item. However, these numbers are very small, making any further interpretation of these findings problematic.

Graph 6.2 Reponses to item *a* with respect to the year of graduation



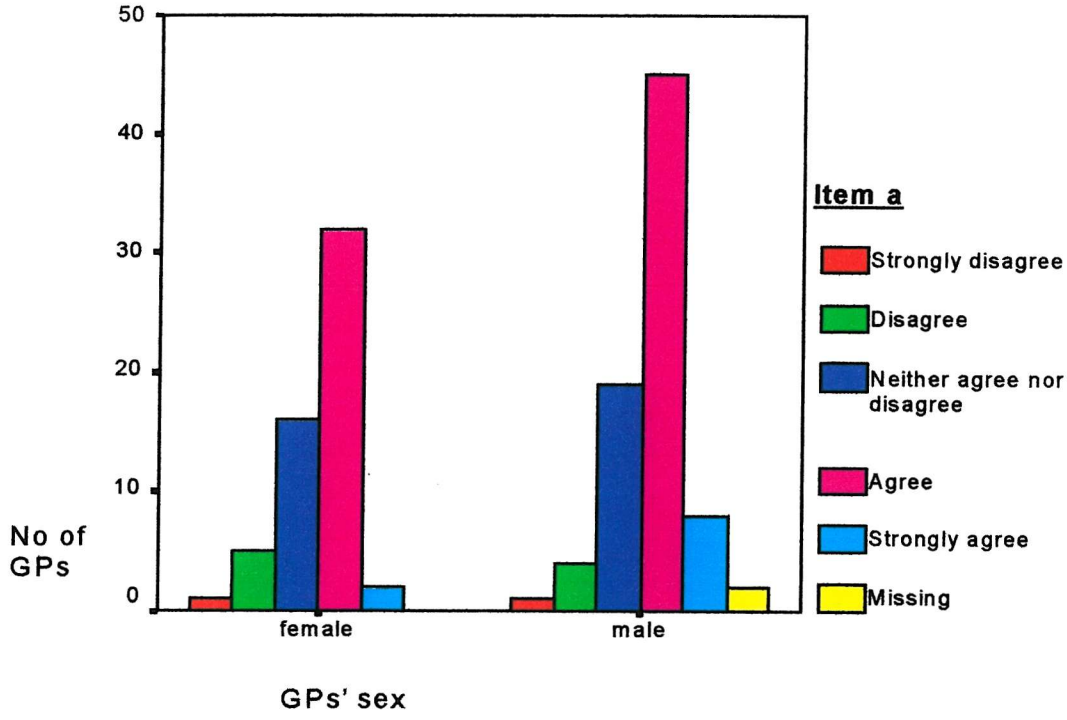
As indicated in table 6.2, the majority of GPs who responded —82% (n=111) graduated between the years 1971 and 1990. Graph 6.2 shows that irrespective of the year of graduation the majority of GPs agreed with item *a.* Looking at graph 6.2 one could suggest that GPs who graduated between 1951-70 were more uncertain about item *a* because a greater proportion of GPs (compared to those who *agreed* for the 1951-1970 period) indicated *neither agree nor disagree* and *don't know.* However, it is difficult to be precise about what this signifies because the observed differences relate to a small number of GPs.

Graph 6.3 Responses to item *a* with respect to practice location



Graph 6.3 suggests that the location of a GPs' practice had little impact on how the GP responded to item *a*. Similarly graph 6.4 below suggests that male and female GPs answered item *a* in a similar fashion.

Graph 6.4 Responses to item *a* with respect to the GP's sex

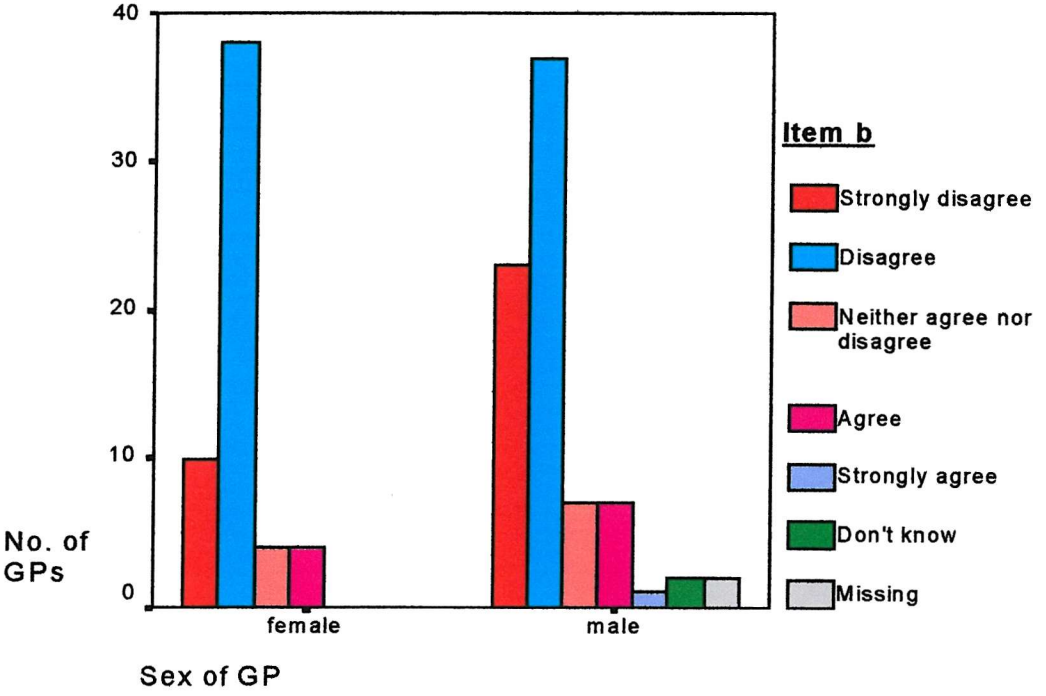


Overall the pattern of responses to item *a* are consistent with the strongly established idea within general practice about family history as a source of social information about individual and family (Royal College of General Practitioners, *The nature of general medical practice* 1996). Given this, it is perhaps surprising that only 88 (65%) GPs *strongly agreed or agreed* with item *a.* One explanation for this is may be the context of the main question which focused on *finding out about a family history of cancer.*

6.7.2 Item *b*---insight into psychological problems

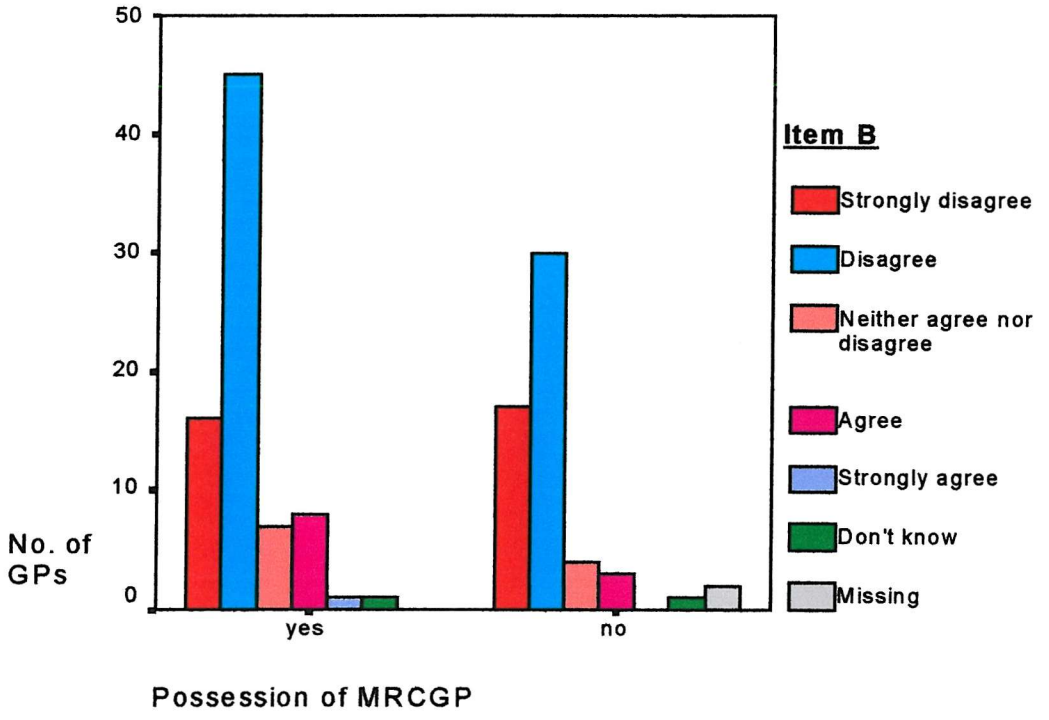
Item *b* produced a high degree consensus—108 (80%): GPs *strongly disagreed or disagreed* that finding out about a family history of cancer would not give insights in to the psychological issues. Again similar patterns of responses were observed irrespective of the GP's sex, MRCGP status, practice location and year of qualification--see graphs 6.5-6.7 below.

Graph 6.5 Response to item *b* with respect to GPs' sex



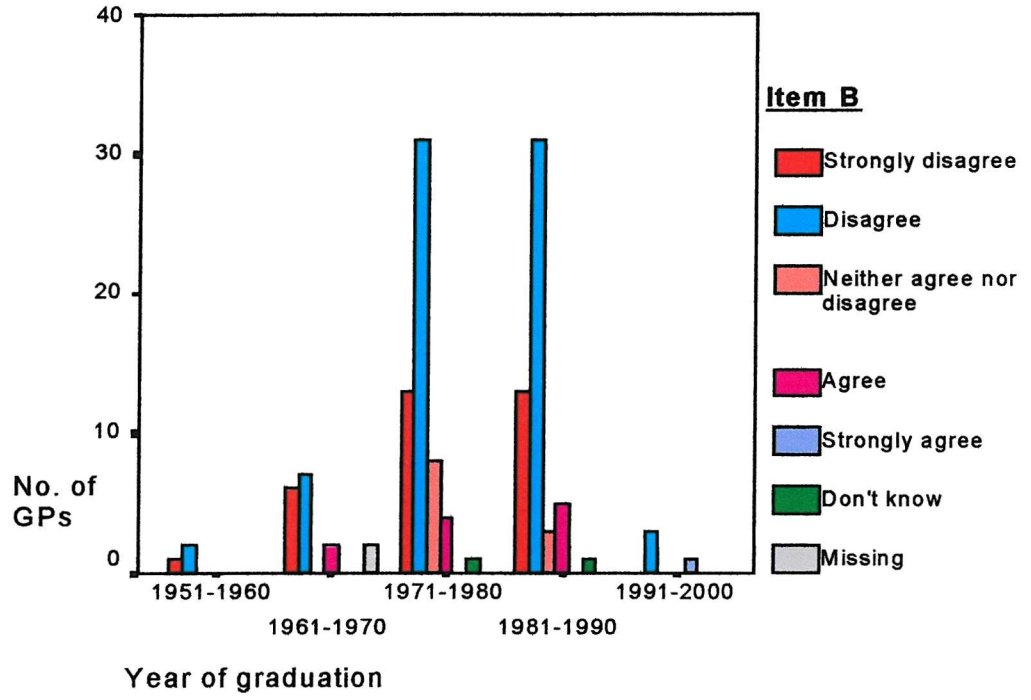
In graph 6.5 the pattern of response was similar between males and females. For example 48/56 of the female GPs (88%) and 60/79 of the male GPs (76%) strongly disagreed or disagreed with the item *b*.

Graph 6.6 Responses to item *b* with respect to MRCGP

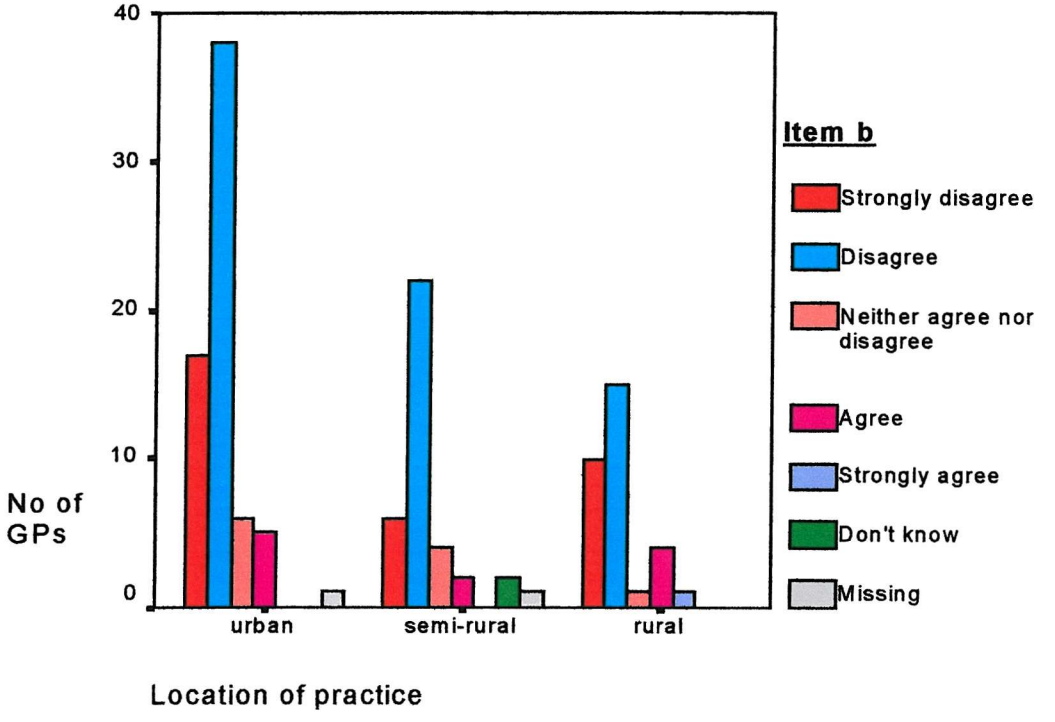


GPs with and without MRCGP responded to item *b* in very similar ways, and this was the case too when responses were examined for the GPs' year of graduation. Again the small variations in the GPs' responses between different years are difficult to interpret, for example GPs in the 1951-60 group showed highest consensus with item *b,* but there were only three GPs in this group.

Graph 6.7 Response to item b with respect to year of graduation



Graph 6.8 Responses to item *b* with respect to location of practice

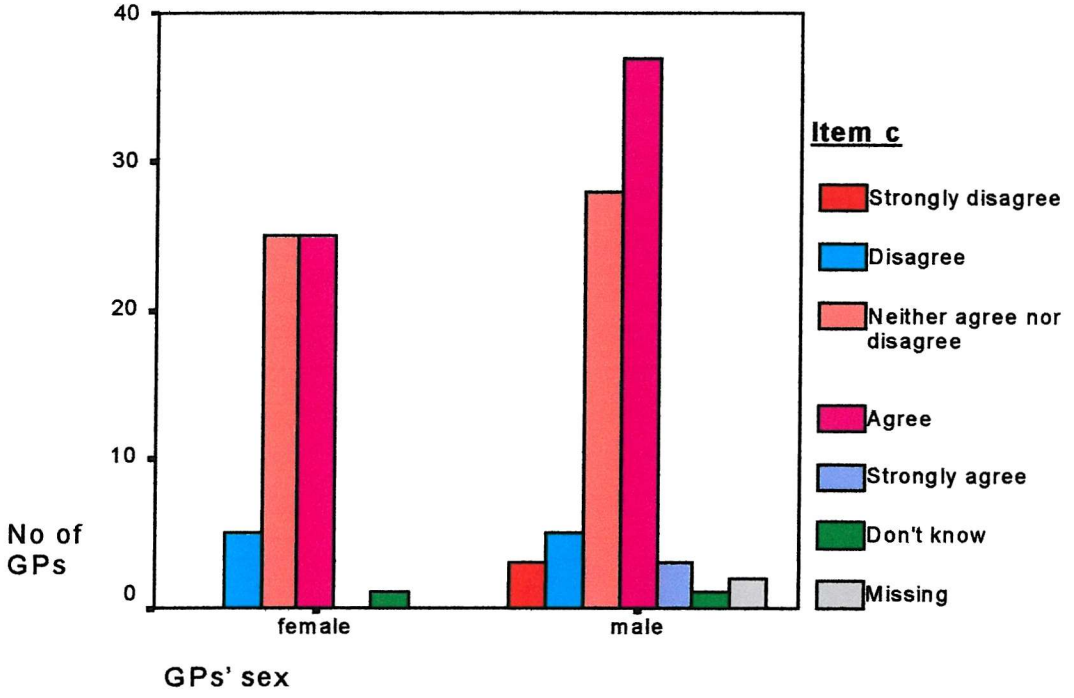


Overall the consistency of responses to item b reflect the strength of the concept of family history as a source of psychological information (Neighbour 1990, RCGP 1996). The level of consensus observed may have been strengthened by the context of the question

which focused on a patient under the age of 50 years being diagnosed with cancer. This context was used to emphasise the possible hereditary nature of the cancer which often present earlier in life. However, this context may have emphasised the inevitability of psychological issues an individual diagnosed with cancer relatively early in life would face. This may also explain why in comparison to item *a* more GPs saw family history as a providing information on psychological rather than social problems.

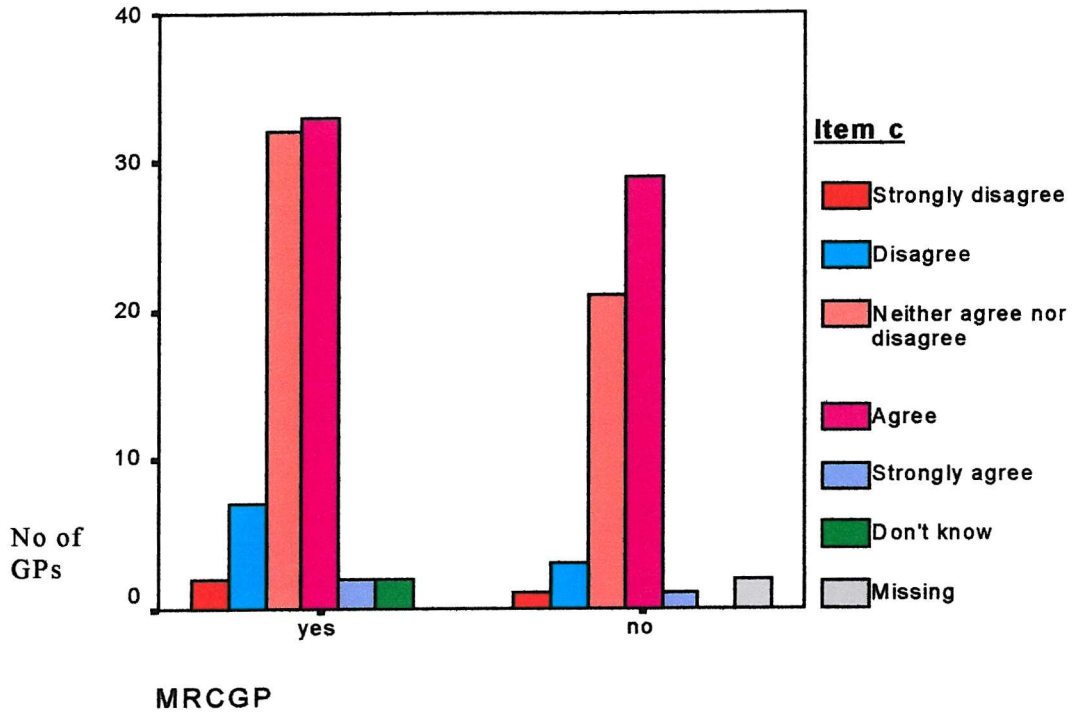
6.7.3 Item *c*---family history explaining why cancer developed

Graph 6.9 Responses to item c with respect to the GP's sex



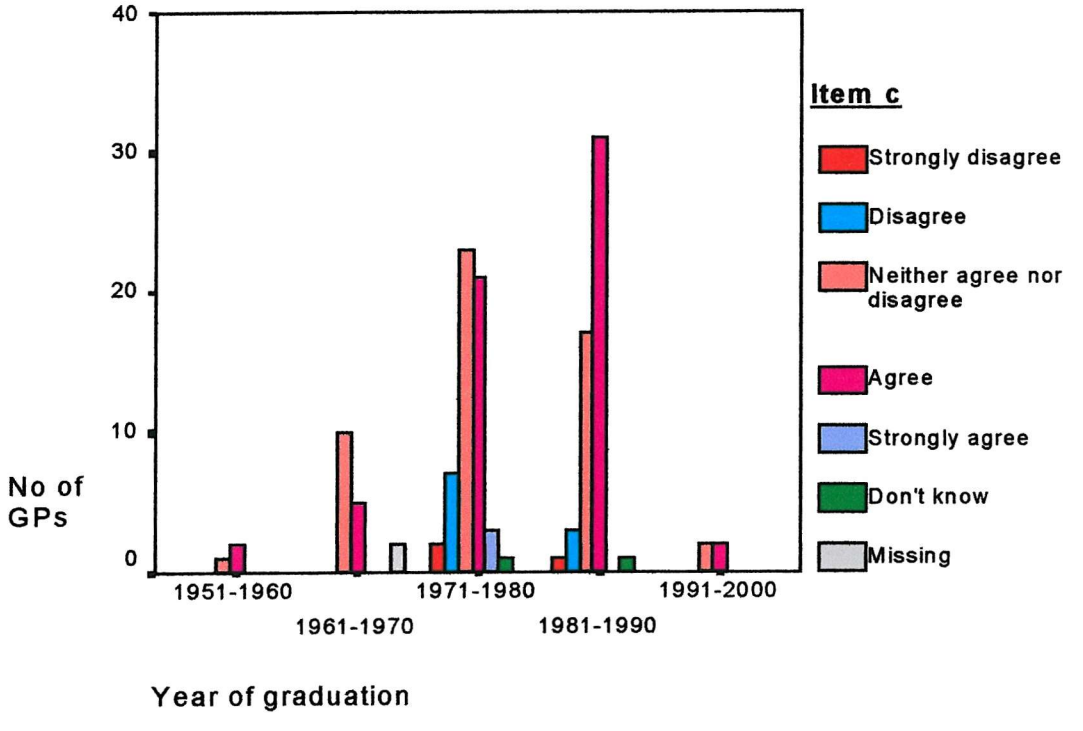
As a proportion of the overall numbers, more female GPs than male GPs were uncertain about a family history providing an explanation for why the cancer had developed compared. Again, the numbers which created this difference (3 GPs) are a small fraction of the total sample.

Graph 7.0 Responses to item c with respect to possession of MRCGP



Similarly, the possession of MRCGP did not produce a difference in how GPs responded to item *c*.

Graph 7.1 Responses to item *c* with respect to year of graduation



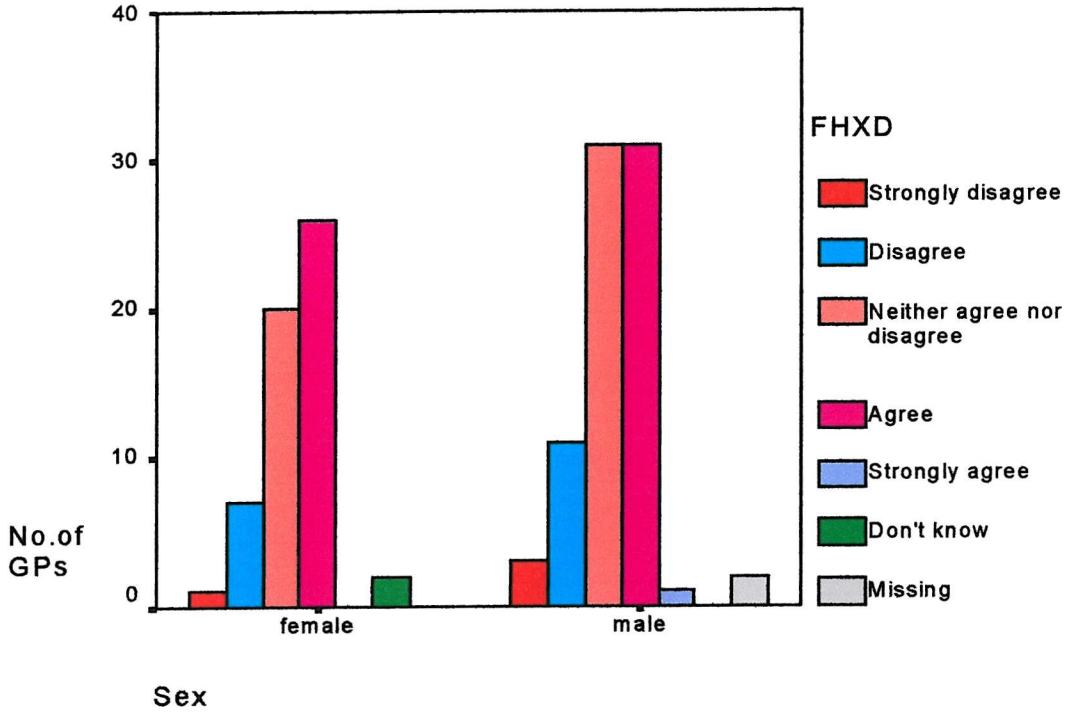
There is an observable trend in the GPs' responses to item *c* when the results are examined by year of graduation. For example, in Graph 7 the proportion of GPs who *strongly agreed/agreed* with item c increased from 1951 to 1990 so that for the first time in the 1981-1990 group GPs who *agreed* out numbered those who were uncertain. The trend was not sustained in the 1991-2000 group but this may be a reflection of their being only four GPs in this group—thus the confidence of this observation is threatened and it is possible that a larger number of GPs in this group would have sustained this trend.

6.7.3 Item *d*---family history giving genetic information

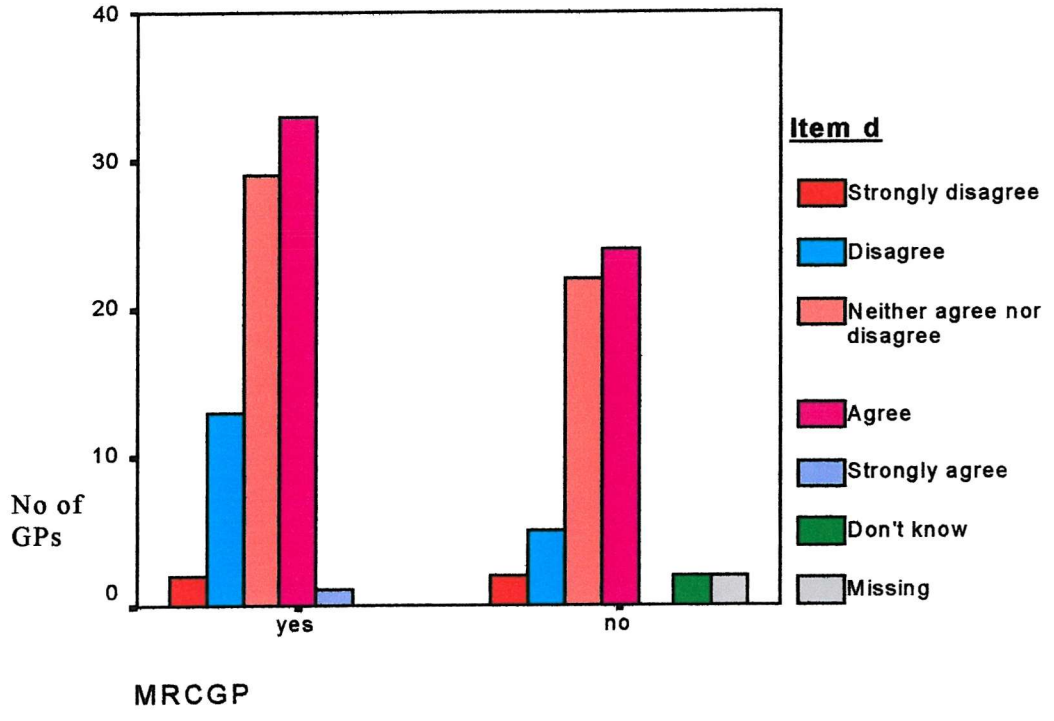
Overall this item created a substantial degree of uncertainty amongst the respondents and caused the largest number of them to *strongly disagree/disagree* in comparison to all the other items. Again the GP's year of graduation is shown to be most influential in determining whether they *strongly agreed/agreed* with item *d*. As for item *c* the proportion of GPs within each group who agreed that family history provided genetic information increased—see graph 7.3. For item *d* the trend continued in to the 1991-2000 group.

Responses to item *c* were similar for male and females with both groups showing higher degrees of uncertainty for item c compared with items *a* and *b*.

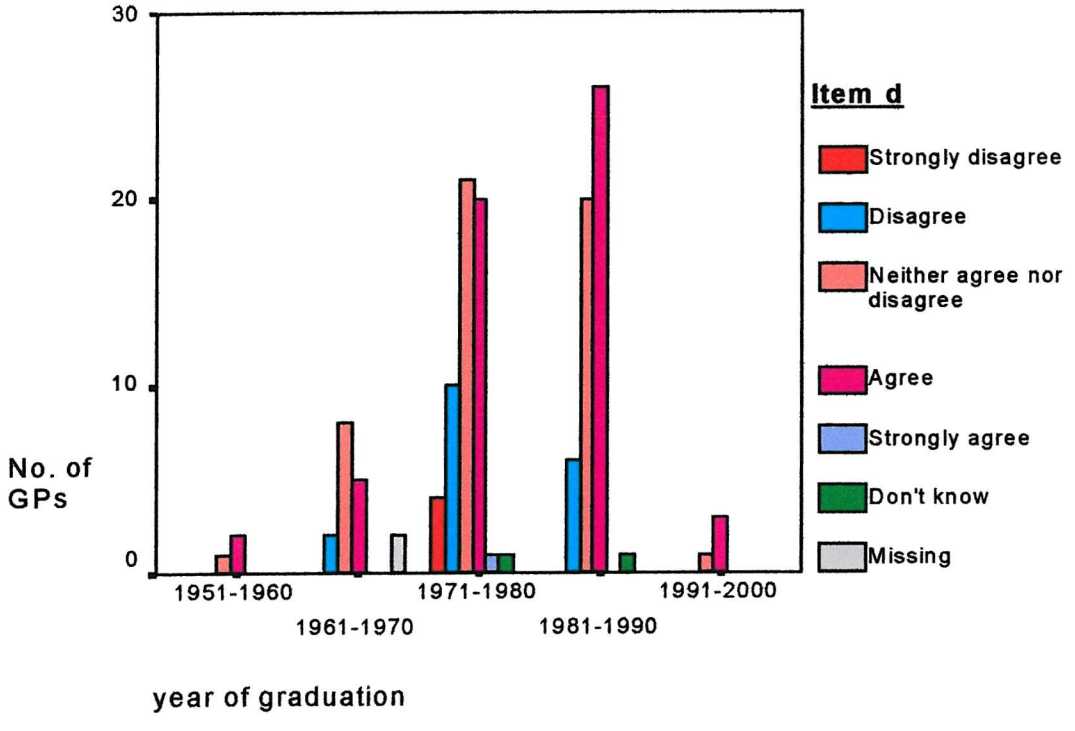
Graph 7.2 Responses to item *d* with respect to GPs' sex



Graph 7.3 Responses to item *d* with respect to MRCGP



Graph 7.4 Response to item d with respect to year of graduation



The GPs responded to items *c* and *d* in similar ways. Both items focused on family history explaining why the cancer had developed with item d emphasising family history as source of genetic information. For item *c,* 55 GPs indicated they *neither agreed nor disagreed* or *didn't know* whether a family history would explain why the patient may have developed the cancer in this context. However, 13 GPs *strongly disagreed or disagreed* with this statement. Overall, GPs were less clear on how family history may explain the occurrence of cancer. This uncertainty was stronger when family history was presented as a source of genetic information in item *d.* In item *d* 53 GPs *neither agreed nor disagreed* or *didn't know* that a family history of cancer would give genetic information about why the cancer developed. Twenty one GPs strongly disagreed or disagreed that a family history would provide them genetic information in the context of why a cancer developed.

6.7.4 Item *e*---family history as information on life-style risks

Responses to item *e* showed a degree of consensus amongst the GPs--in that a family history would give information about a family's life-style risks that may explain the occurrence of the cancer.

6.7.5 Item f ---family history will allow me to assess if others family members are at risk

This produced the greatest degree of consensus between GPs when 114 (84%) *strongly agreed or agreed* that finding out about family history of cancer would allow them to assess if other family members were at genetic risk of developing cancer. This was in contrast to item *d* where only 58 GPs *strongly agreed/agreed* that a family history provides genetic information about why the cancer developed. However, 10 GPs strongly disagreed /disagreed—the lowest number expressing dissent for all of the items. Again, for items *c,d,e,*and *f* GPs' responses did

not appear to be influenced by sex, possession of MRCGP, year of qualification or the practice location.

6.8 Factor analysis

In addition to the descriptive statistical analysis a factor analysis of the questionnaire data was undertaken to test the hypotheses derived from the qualitative data. The factor analysis is presented below.

6.8.1 The aims of factor analysis:

1) To determine how the categories/measures deployed in the questionnaire items *a-f* co-varied with one another.
2) Whether the pattern of co-variance revealed by the factor analysis followed the pattern identified in the qualitative study (see below).
3) To test the construct validity of the hypothesis constructed from the qualitative study.

6.8.2 Factor analysis methodology

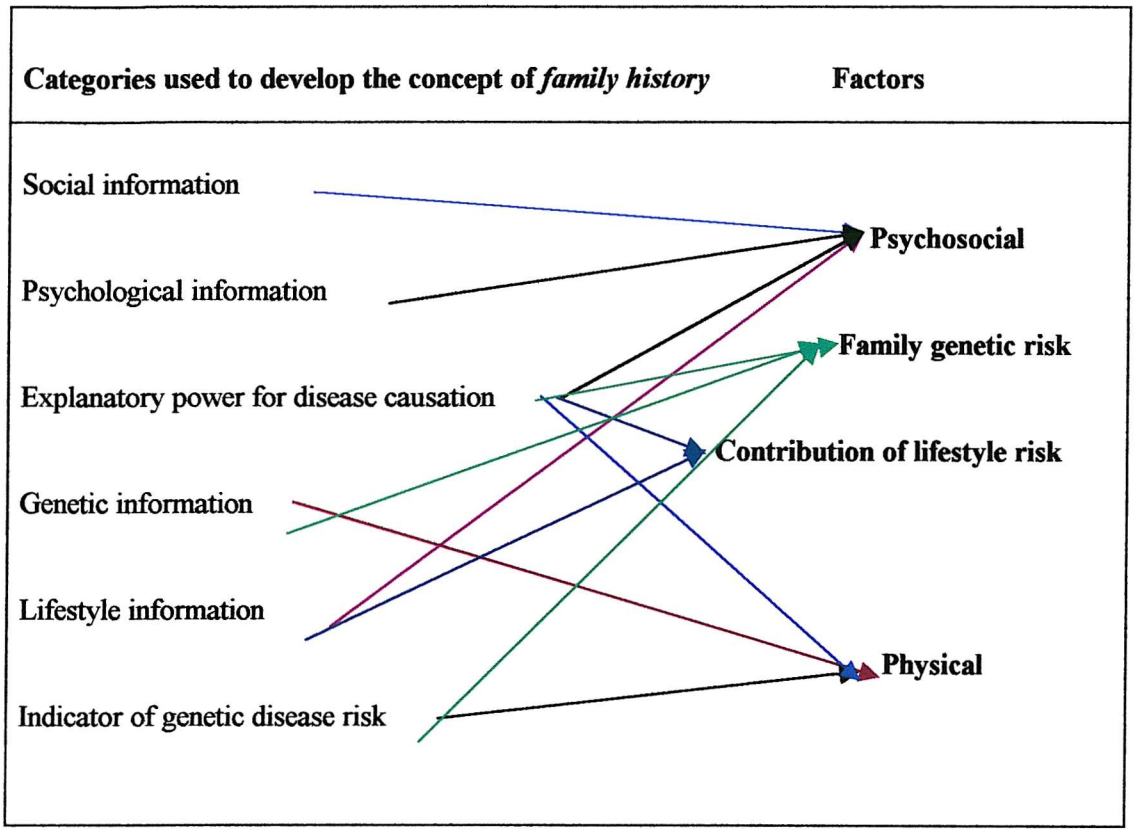
Factor analysis is an analytical technique for identifying *factors* that statistically explain variation and co- variation among a set of variables, attributes, responses or observations (Kinnear and Gray 1994). Generally, the number of factors is considerably smaller than the number of measures and, consequently, the factors are considered a succinct representation of the measures. Thus, factor analysis is often termed a data reduction technique because it reduces a large number of overlapping measured variables to a much smaller set of factors (Green, Salkind and Akey 1997). In studies where different

sets of measures reflect different dimensions of a broader conceptual system (e.g. the categories/measures deployed in the GP questionnaire), factor analysis can produce factors that represent these dimensions. More specifically, the factors can correspond to constructs (i.e. unobservable latent variables) of a theory that helps to understand behaviour.

Ideally the variables to be analysed should be quantitative, have a wide range of scores and be uni-modally, symmetrically distributed. However, factor analysis can be applied to variables with a more limited range of scores as found in a Likert scale (Green, Salkind and Akey 1997).

Before presenting the factor analysis, it is useful for reasons of clarity, to discuss how the terms used in factor analysis map onto those used in the qualitative study, and the GP questionnaire. Each of the categories (appearing in items a-f) represent a specific dimension of the concept/construct ***family history,*** which in turn is one concept among several (see above) contributing to produce the theme-- ***genetics in the generalist context*** (see chapter four). The theme, genetics in the generalist context is the ***broad conceptual system*** referred to in factor analysis--a complex theoretical abstraction that represents a range of ideas, processes and actions described by GPs when they spoke about genetic advances, generalism and their personal clinical practice. The *factors* referred to in factorial analysis are akin to the categories in the qualitative analysis. Figure 6.3, is a representation of the relationship between *factors* and the categories.

Figure 6.3 Qualitative categories and the factors used in the factor analysis



The categories shown on the left hand of the figure 6.3 were operationalised in the questionnaire items (a to f).

6.8.3 Applying factor analysis

Before carrying out factor analysis the data had to meet specific requirements (Green, Salkind and Akey 1997): -a) that the variables to be analysed be quantitative and have a range of scores—the qualitative categories had been deployed in the GP questionnaire and were measured by a scale thus rendering them quantitative.

b) that the factors in factor analysis represent a set of measures succinctly. I was confident that the categories (measures when operationalised in the questionnaire) from

which factors were developed were robust for the reasons given in chapters five and so the factors would be reliable.

Before the factor analysis could be performed the questionnaire data had to be manipulated which was done using *Stata* a computerised statistical package. This part of the analysis was overseen by Professor Paul Little in the department of Primary Medical Care in Southampton who has substantial expertise in this area based on a theoretical understanding of the procedure, and experience in its application. The specific procedures Professor Little contributed to were factor extraction and factor rotation. The primary objective of factor extraction is to make an initial decision about the number of factors underlying a set of measured variables. Factor rotation has two goals: 1) to statistically manipulate the results to make the factors more interpretable and 2) to make decisions about the number of underlying factors.

Ideally, in factor analysis four or more measures should be chosen to represent the construct/hypothesis. In the GP questionnaire the construct/concept of family history was represented by six categories/variables. From these four factors were generated to represent the construct by collapsing categories/variables shown in figure 6.3.

6.8.4 The main construct/hypothesis:

In the context of a patient developing cancer under the age of 50 years, GPs collect family history to assess risks associated with life-style, the psycho-social impact of being diagnosed with cancer and not to collect genetic information.

The construct/hypothesis was identified by the qualitative study (see section 6.2.2). As described in chapter four, during the process of qualitative analysis, issues and categories are identified which are then linked in different ways to produce concepts and themes. This process of identification followed by linkage creates patterns in which specific categories co-vary i.e. are present together (positive co-variation) or mutually exclusive (negative co-variation) in a specific context. Thus, the qualitative study suggested a pattern for how the categories operationalised in the GP questionnaire might co-vary, and by extension the *factors* derived from them (see figure 6.3). Further, accepting that qualitative findings are transferable (Bryman 1988) to GPs whose contexts are similar to the GPs interviewed in the qualitative study, it would follow that if such were interviewed we would identify similar if not the same categories and patterns of variation described by the qualitative study—indeed, this is born-out by the results of subsequent qualitative studies of GPs on the topic of the new genetics (Watson et al 1999 and Elwyn et al 2000). Additionally, a representative sample of GPs responding to a questionnaire, in which categories from a qualitative study are operationalised, may be expected to respond in a manner that reflects the pattern of variation suggested by the qualitative results. This assumption carries weight if one accepts Glaser's view (1978, 1992) that grounded theory analysis has a *predictive* function because of the level of theoretical abstraction achieved by constant comparison analysis, which makes the findings *generalisable*. Assuming this to hold true in the context of the GP survey then the pattern of response to be expected is represented in table 6.5 below.

Table 6.5 : Pattern of responses to questionnaire options as predicted by the qualitative study.

Categories in the questionnaire items risk	**a** social	**b** psychological	**c** explanation	**d** genetic info.	**e** lifestyle risks	**f** genetic
a social		++	-	--	+	--
b psychological	++		-	--	+	--
c explanation	-	-		++	++	++
d genetic info.	--	--	++		--	++
e lifestyle risks	+	+	++	--		--
f genetic risk	--	--	++	++	--	

++ indicates a strong likelihood of these categories occurring together and + indicates categories are likely to occur together.
-- indicates the categories are strongly mutually exclusive, – indicates the categories are weakly mutually exclusive.

Option ***a*** is used to illustrate the pattern of co-variation between categories in the table. In the context of a patient under the age of 50 being diagnosed with cancer, a GP who takes a family history to explore the social problems that a patient would face as a consequence of the diagosis would be expected to explore the psychological impact too. Further, a GP who explores social and psychological factors is more likely to consider the effect of lifestyle factors on the development of cancer. However, the same GP would be unlikely to use family history to clarify genetic risk for the patient and the family.

6.8.5 The factor analysis

Table 6.6 Spearman's Rho

Correlation Coefficient (sig. 2-tailed). N=133

	Psycho-social	Physical	FHxE	FHxF
Psycho-social	1.00	-0.008	**0.255****	0.175*
		0.925	0.003	0.044
Physical	-0.008	1.000	0.110	0.176*
	0.925		0.208	0.043
FhxE	**0.255****	0.110	1.000	0.059
	0.003	0.208		0.501
FHxF	0.175*	0.176*	0.059	1.000
	0.044	0.043	0.501	

** Correlation is significant at the 0.01 level (2-tailed), * correlation is significant at the 0.05 level. The figures in red are the *p-values.*

In the table above, +1 signifies complete correlation and –1 complete negative correlation

Psycho-social = item *a* (responses which strongly agree or agree) + item *b* (disagree and strongly disagree)

Physical = item *c* (strongly agree or agree) + item *d* (strongly agree or agree) + item *f* (strongly agree + agree)

FHxE = item e, family history as information of life-style factors to explain occurrence of cancer

FHxF = item f, family history allows assessment of genetic risk to other family members

The items a, b, c, d, e and f are questionnaire items see appendix two.

On inspecting the matrix, the following observations appear to be have the greatest level of inter-correlations:

1) **Psycho-social and FhxE, (lifestyle category)**—this correlation was the strongest and echoes the pattern found by the qualitative study. GPs who collected a family

history of cancer to assess the social and psychological consequences of the disease to the patient and their family were more likely to collect life-style information to gauge the possible exposure to shared carcinogens e.g. smoking. In accordance with the qualitative hypothesis co-variation between these factors suggests the GP is prioritising a holistic approach to understanding the occurrence and the consequences of the cancer. It is possible that GPs whose responses show these correlations are either unaware of genetic advances or like the informed GPs in the qualitative study are wary of the limitations of genetic advances in predicting risk and the ethical issues associated with it.

2) **Physical and FHxF** showed a weaker correlation (family history used to assess genetic risk to others). Responses which showed this correlation suggest these GPs understood family history as genetic information, which could explain why the cancer had developed. Additionally, these GPs appeared to understand the role of family history in assessing the risk to other family members of developing the cancer.
3) **Psycho-social and FHxF** showed the weakest correlation which suggests that when family history is used to provide psychological and social insights it is less likely to be used as genetic information--this supports the qualitative hypothesis.
4) **Psycho-social and physical** showed the least degree of correlation. GPs who collected family history to evaluate the social and psychological consequences of cancer for the individual and their family were least likely to use family history to assess the genetic risk to others or understand family history as a providing genetic information about why the cancer had developed.

When taken as a whole the correlations produced by the factor analysis closely resemble the pattern suggested by the qualitative study and support the overall hypothesis (see table 6.4 above). It is important to emphasise that it is the overall correlation/pattern suggested by the factor analysis which is used to make the final interpretation of whether the hypothesis is supported and not any single correlation. Thus overall, the hypothesis generated by qualitative study is supported by the factor analysis of the questionnaire data. The next chapter will discuss the findings of both the qualitative and quantitative studies further in relation to each other and to the work published in this area since these studies were completed.

6.9 Summary

In this chapter I presented the quantitative part of the thesis—the GP survey which used a postal questionnaire developed from the qualitative data. The survey examined whether the hypotheses generated by the qualitative data would be upheld by a larger random sample of GPs. To examine the integrity of the hypothesis generated from the qualitative data I undertook a factor analysis of the survey results to establish if the categories deployed in the questionnaire co-varied as suggested by the hypotheses generated from the qualitative study. The survey up-held the findings of the qualitative study, and the factors analysis echoed the variation between categories suggested by the qualitative analysis.

In the next chapter I will discuss the findings of both studies.

Chapter seven

Discussion and conclusions

7.1 Introduction

The discovery of genes conferring susceptibility to common conditions was rapidly followed by a rhetoric promising the development of predictive genetic tests, and improved diagnostic and therapeutic technologies, which would lessen the morbidity and mortality resulting from common conditions (Genetics Research Advisory Group 1995a, MacIntyre 1997, Bell 1997). It was assumed that the general public, once aware of genetic advances would demand access to services in order to clarify their own, and their family's risks. At the same time, it was evident that existing regional genetic services would be unable to cope with any increase in referrals from primary care (Harris 1992). Thus, the promised applications of molecular genetics in the prediction and diagnosis of disease was seen as an opportunity for the rational development of clinical services in the NHS (Kinmonth et al 1999). Starting in the early 1990s, and continuing to the present, clinical geneticists, GPs, policy-makers and government (Harris 1992, Qureshi and Reaburn 1993, Austoker 1994, Calman Officer 1995, Gill and Richards 1998, RCGP, *Genetics in Primary Care* 1999) all identified primary care as pivotal to managing people's concerns about developing inherited common diseases such as breast cancer. General practitioners were specifically identified as requiring education and training in the *new genetics* (Calman 1995, Genetics Research Advisory Group 1995a, Harris and Harris 1995). One view of what education and training for GPs would entail was made explicit by Austoker

(1994) in defining the future role of GPs in the genetic testing for cancer, see boxed text below:

Box 7.1—Roles defined for GPs

- An awareness of the genetic dimension to common diseases such as cancers.
- The dimensions of family history that are needed to assess genetic risk e.g. age of onset of the condition in the relative, the degree of relatedness etc.
- The scope of genetic testing for common diseases e.g. it benefits, limitations and risks
- Issues surrounding pre and post test counselling and continuing support for those at high risk
- Ability to explain the meaning of testing positive and negative
- Explaining implications for other family members
- Knowing who should be referred for a specialist opinion
- Knowing where to refer patients to
- Knowing what post test counselling will be required
- What advice can be offered to those who do not require referral
- Issues around recording sensitive information in notes/ ownership of genetic information.

The studies described in this thesis set out to examine the GP's perceptions, ideas and opinions of the role(s) being defined for them by experts. This chapter provides space to reflect upon GPs' responses made in the qualitative and quantitative studies, and to develop a more theorised discussion drawing on data and literature.

The chapter is divided in to three sections: in the first section I discuss the results of the qualitative and quantitative studies, in relation to the findings of studies published after the work in this thesis was completed. In the second section I develop a more theoretical discussion of the findings by examining genetic advances, general practitioners, and generalism through a more anthropological lens: viewing clinical medicine and general practice as a social and cultural practice. Finally, in section three, I will present my conclusions and reflections on future research possibilities.

7.1.1 Section one—the findings of the qualitative study

Reserved support. In general, informed GPs interviewed in the qualitative study were positive about the clarification genetic research was providing about the genetic basis of common conditions. GPs acknowledged they would have a role to play in managing peoples' enquiries about genetic risk to common conditions such as breast cancer. They looked forward to the promise of better screening, diagnostic and therapeutic technologies and wanted to learn more about the subject. These general findings are echoed in research by others exploring GPs opinions and perceptions of their role in assessing genetic susceptibility to common conditions (Emery, Watson, Rose and Andermann 1999, Fry et al 1999 and Elwyn, Iredale and Gray 2000).

However, a more detailed look at the findings of the qualitative study showed there were differences of opinion between GPs, which were influenced by how well they were informed about genetic advances. Informed GPs were reluctant to identify patients with genetic susceptibility to breast cancer, and other common cancers for a

number of reasons including uncertainty about the stability of *new genetic* knowledge and related innovations such as genetics testing, which are detailed in the category: *instability of new genetic knowledge.* These findings resonate with Michie et al's (1996) grounded theory study which aimed to understand patients' responses to predictive genetic testing in the context of familial adenomatous polyposis (FAP, a late onset genetic condition). They identified professional uncertainty among surgeons and nurses about genetic testing for bowel cancer, which was in contrast to their certainty about established bowel screening tests e.g. sigmoidoscopy. They saw conventional tests as providing concrete, categorical, reliable and immediate results, unlike genetic tests which were seen as research, abstract, probabilistic, and delayed. One criticism of the Michie et al's category of *professional uncertainty* was its derivation from patients' interviews in which they alluded to professionals' responses to genetic and mechanical bowel screening strategies. Thus, in the absence of the interviews with health professionals the integrity and credibility of this category is open to question. Nonetheless, the category of professional uncertainty was supported by the GP study in this thesis.

Reluctance in raising the issue of a family history of breast cancer. Thus, on the one hand, GPs were supportive and positive about the genetic advances being made, whilst on the other, they were cautious about potential negative consequences for themselves and patients. This apparently contradictory stance makes sense when it is understood as GPs' responses to knowledge about the potential negative impact of genetic advances and not a consequence of inconsistency or ignorance. In this respect GPs echo lay responses as captured by Jallinoja et al's (1998) survey of 1169 people, which aimed to evaluate attitudes towards genetic testing in Finland. They found

people adopted contradictory responses when faced with the moral and ethical issues raised by genetic testing and suggested that health professionals would need to help their clients cope with their contradictory feelings and so help them find acceptable solutions for their specific problems. However, health professionals may be unable to perform this role as explained by the category *ethical dilemmas and the therapeutic gap,* which described GPs using ethical objections to identifying people with genetic susceptibility in the absence of effective screening, and therapeutic technologies. For example, in the context of breast cancer where the only intervention available to women was to undergo a bilateral mastectomy, then GPs opted not to raise the issue of risk even when a woman's family revealed her to be at high risk. These doctors framed their reticence to discuss genetic risk as 'ethical,' a use of the term which appears to refer to making decisions on the patient's behalf and which may itself be considered by some as unethical. Suchard et al's (1999) questionnaire survey of GPs' views on genetic screening for common diseases also found ethical issues such as increased anxiety among patients, screening for untreatable diseases, potential for insurance discrimination, and disclosing information among family members deterred GPs from identifying women with genetic susceptibility to breast cancer. Further support for ethical concerns acting as deterrent is provided by Watson et al (1999). They carried out a qualitative study of GPs in Oxfordshire and found the absence of effective screening and therapeutic technologies stopped GPs raising the issue of family history and genetic risk. Thus there is evidence from both qualitative and quantitative studies which support the category *ethical dilemmas and the therapeutic gap.*

Informed GPs' ethical concerns were reasonable given that many were aware of the lack of effectiveness of mammographic screening for younger women with a family history of breast cancer (Kerlikkowske 1996), unproven benefit from chemo-prevention, unsuitability of tamoxifen for women of child bearing age, failure of prophylactic bilateral mastectomy for carriers of BRCA1 to remove all the breast tissue (even with the most radical operation) (Eeles 1996, Lerner 2001).

GPs reluctance to raise the issue of a family history of breast cancer is supported by two recent quantitative studies of general practice. The first was carried out in Cambridgeshire by the Womens' Concerns Group (2001) which found the issue of a family history of breast cancer was raised by women in only 5 consultations per 1000—an average of 0.6 per physician per month. However, GPs raised the issue 6.6 times more often than women. The group estimated that for each 1000 women, aged 16 years or older, about 15 a year will raise the issue of a family history of breast cancer with their GP. Similar results were found by DeBock (2001) for women consulting their GPs about a family history of breast cancer in a general practice in Leiden, in the Netherlands. Although the design and sampling of these studies limits the representativeness and generalisability of their findings, they do suggest some change in GPs readiness to raise the issue of genetic risk may have occurred since the qualitative study in this thesis was completed. However, another qualitative study of Oxfordshire GPs' views on their role and educational needs (Watson et al 1999) found GPs thought consultations on family history and genetic conditions were uncommon, and that GPs were reluctant to discuss family history of breast cancer due to ethical objections.

What did GPs know about clinical genetics? GPs knowledge of genetics was strongest in relation single gene disorders which followed a Mendelian pattern (e.g. cystic fibrosis), and chromosomal disorders (e.g. Down's syndrome). In the context of these types of diseases GPs identified a clear role for themselves. They reported using family histories collected during pre-conceptual, antenatal and child health surveillance clinics and in ordinary consultations to identify genetic risk and then refer to a clinical geneticist or obstetrician. Doctors who were uninformed about genetic advances in the context of common late onset conditions (e.g. breast cancer) envisaged genetic advances would impact most on established genetic diseases such as cystic fibrosis. All GPs, irrespective of whether they had attended the course thought they did not possess the skills or knowledge required to take a family history in order to assess genetic risk in the context of common conditions.

Fry et al's (1999) survey of GPs showed they experienced difficulty in assessing genetic risk, with a tendency to over-estimate the risk. They found GPs' willingness to identify genetic risk was associated with a younger age (<40 years), training in genetics, attending continuing medical education and involvement in obstetrics. On the whole these findings were supported by the GPs interviewed in the qualitative study: only one GP with close links to a regional genetics department was whole-heartedly willing to identify people at genetic risk. Another survey of GPs by Mennie et al (1998) showed that GPs who were clinically involved with Huntington's disease were more likely to know about pre-symptomatic testing and to know which of their other patients were at risk. This was not up-held by three GPs who looked after patients with Huntington's disease.

GPs' idea of their role. GPs saw their role along traditional lines as gatekeepers, referring to specialists for expert analysis of family histories of cancer, themselves providing continuing support and managing the patient's and the family's risk in the context of their psychosocial lives. Emery et al (1999) found that GPs believed they have a role to play in identifying people with genetic susceptibility to common diseases. Surveys by Freidman (1997), Suchard (1999) and Fry et al (1999) found there was general support for the principle of genetic counselling and/or screening for a family history of cancer. GPs believed their role would involve taking detailed family histories and performing a gate-keeping function. However they found that only between 27% and 39% of GPs were confident in performing this role. Unfortunately, none of these studies were able to explore in any depth why so many GPs lacked confidence. The findings of my qualitative study suggest that GPs confidence is related to their idea that genetic risk assessment for common conditions is complex and is made more difficult by alerting patients to the social, ethical and psychological consequences of their genetic risks. Only one study, from the United states (O' Malley et al 1997) conducted soon after the identification of BRCA 1, has shown family practitioners prepared to offer BRCA 1 testing and provide advice to mutation carriers. The limitation of this study is that there was no clarification what this advice would be, and whether family practitioners were aware of the limitations of the BRCA 1 testing. It is probable these family physicians were responding to the highly optimistic debates on BRCA 1 screening at the time, which largely ignored the limitations of BRCA 1 testing.

Collecting family history. GPs in the qualitative study did not describe collecting a family history of cancer in any systematic way. Most often GPs described recording a

general family history of common conditions (usually excluding cancer) only when the patient first registers and then in a cursory fashion. This information was rarely reviewed, recalled or linked to other family members. GPs were unclear of the extent and depth of the family history required to assess genetic risk for breast cancer, for example: the number of first and second degree relatives affected, distinguishing between maternal and paternal relatives, the age of onset of breast cancer in other members, whether the breast cancer was bilateral, recording other cancers or conditions that were associated with genetic susceptibility e.g. prostate cancer in male relatives, ovarian cancer, endometrial cancer and so on. These findings were echoed by Watson et al's (1999) qualitative study. However, Summerton's and Garrod's (1997), questionnaire study to 291 GP (response rate of 177/291= 61%) found that 84/177= 48% said they recorded family history of breast cancer at registration. Other conditions recorded were ischaemic heart disease 94%, and colorectal cancer 31%.

More recently, a health centre in Cambridgeshire sent out a questionnaire to 2 265 patients between the ages of 35-64 requesting they provide a family history of cancer (Leggatt et al 1999). The aim was identify patients at increased risk of breast or colorectal cancer. Only 920 (40.6%) agreed to take part from which 29 patients were assessed to be at significantly increased risk, and 11 were of such high risk they were offered screening after counselling from a clinical geneticist. In this study the single handed GP received support from a consultant geneticist in assessing a patient's family history for risk. This GP was committed to being proactive in identifying family histories of cancer among her patients. This commitment to surveillance appeared to transcend any concerns for the negative impact of the questionnaire on her patients. At no point in this paper were reasons for patients' non-response

addressed. The psychological, and social impact of the questionnaire were completely ignored. This was surveillance without regard for patients who may have been unable to collect family histories of cancer, or discussion of the management of patients of moderately increased risk but insufficient to justify screening or further surveillance. The high non-response to this questionnaire is probably indicative of the concerns and reservations patients had about receiving it. Only the GP I interviewed as part of the theoretical sample because of her experience in a community based cystic fibrosis screening programme, advocated GPs taking such a proactive role.

Work-load implications for general practitioners. Most GPs thought that collecting family histories, assessing them, and then advising the patient about their risks and need for further assessment would be a low priority because of opportunity costs. GPs recognised that once patients were alerted to the implications of a family history of cancer, some would require continuing psychological and emotional support. However, all the GPs interviewed could not estimate how many patients on their list were likely to have a family history of cancer but most thought the number would be high because cancer is a common condition. Clarification on this point is offered by Johnson et al (1995). Based on their study of 8 109 people attending for health checks, they estimated that 40 to 50 patients aged between 35 and 64 years in a general practice population of 2000 will have a "certain family history" of one of the four common cancers in a first degree relative. However, what is still unclear is how many of these people will attend their GPs with concerns about their family history of cancer. The findings of the qualitative study suggest that this number was low at the time of interview. Since then a study suggests there will 15 consultations per year per

average list for a family history of breast cancer (Women's concerns study group 2001).

The way forward GPs in the qualitative study hoped links would be developed between specialist genetic centres and general practices. They favoured a model of service delivery in which genetic nurse specialists, facilitators, or clinical geneticists would provide an out-reach service to groups of practices. GPs thought this service would review patients' family history of cancers and allocate a risk on which further treatment decisions, and support would be based.

7.2 The results of the quantitative study

The quantitative study was a survey of random sample of 200 GPs in Hampshire. Of the original sample, 10 GPs had retired, moved or died leaving 190 GPs able to respond of which 135 did giving a response rate of 71%. The aim of the GP questionnaire was to enhance the understanding gained by the qualitative study and not to generate a statistical analysis of great analytical precision or predictive power. The methodological issues raised by combining qualitative and quantitative studies has been examined in detail in chapters three and six.

The questionnaire examined one of the hypothesis generated from the qualitative study. **In the context of breast cancer GPs collect family history to obtain information on psychological, social and life-style information more than genetic information.** This hypothesis was upheld by the survey data. GPs were asked why finding out about a family history might be useful when a patient under the age of 50

years was diagnosed with cancer. Altogether 108/135 = 80% agreed it was to find out about the psychological problems, 114/135=84% agreed it would allow them to assess risk to other family members. However, only 58/135=43% agreed a family history would provide them with genetic information, and 65/135= 48% agreed it would explain why the cancer developed. GP's sex, possession of MRCGP, location of practice, whether it was a training practice did not affect how they responded. There was some indication that GPs more recently qualified were more likely to agree with the idea that family history provides genetic information. However the numbers of GPs who qualified from 1990s onward were so limited in the sample that no firm conclusion could be drawn from this. Other researchers (Fry et al 1999, and Watson et al 1999) also detected similar trends.

The factor analysis of the questionnaire data added further support to the findings of the qualitative study. The qualitative study suggested that GPs who collect family history to assess the psycho-social impact of a cancer in a family were less likely to collect details to allow its use as a genetic tool. The factor analysis showed a high correlation between family history used to assess psychological and social information and life-style risk factors. When this was the case there was a negative correlation for family history as genetic information.

7.3 Section 2: Theoretical discussion of the results

In this section I have drawn on the findings of the research described in this work to inform a more reflective and theoretical discussion of how genetic advances impact

on general practitioners and the idea of generalism. In constructing my discussion I have drawn on literature from medicine, anthropology and sociology.

General practice has been identified by policy-makers, the Royal College of General Practitioners (RCGP), primary care experts and clinical geneticists as occupying a pivotal position for the future application of genetic advances to society, (Harris 1995, House of Commons Report 1995a, RCGP 1999, *Genetics in Primary Care,* Kinmonth, Reinhard, Bobrow and Pauker 1999). As the major point of first contact with the NHS, general practice will be the place where the majority of enquiries about genetics occur. The recognition of general practice as pivotal to implementing genetic advances reflects concerns about meeting the expected increase in demand for genetic counselling which can not be met by existing genetic services: the ratio of GPs to consultant geneticists is approximately 500 to one.

It has been assumed that GPs will extend their existing services to include providing patients and families with information, advice and genetic risk assessment for common conditions like breast cancer (Austoker 1994, and RCGP 1999). The delegation of these tasks comes at a time when GPs perceive they are being asked to take on more and more with the consequent marginalisation of *"the real substance of their work"* (Heath 1995). However, based on the findings of my qualitative and quantitative studies I shall argue it is GPs skills, knowledge, and attitudes associated with carrying out the *real substance*: patient centredness, patient advocacy that makes general practitioners well suited to managing the communication of genetic risk, and the psychological, social and ethical issues raised by genetic advances.

I argue that the presentation of the genetic advances as a revolution generating new tasks for general practice detracts attention from its real potential to preserve our traditional skills that are (perceived) to be under threat. The rhetoric of revolution inherent in the epithet "new genetics" obscures the extent to which the implementation of new genetic evidence in general practice will need to draw on both "traditional" as well as "new" skills and knowledge. I will also discuss how advances in genetic medicine might disrupt the recent shifts in power from experts and general practitioners to patients.

Since the 1990s, general practice has emerged as one of the most contested disciplines in the National Health Service. Agencies such as the governments, policy-makers, the RCGP, patient-groups and secondary care professionals have tended to look upon primary care simply as an object to be fashioned in order that such agencies can exercise greater control and influence over its functions and future development. The 1990 contract was imposed on general practitioners with the aim of increasing efficiency, effectiveness, choice, quality, and accountability in general practice (Lewis 1997). This was presented to the public and the medical profession as an act of modernisation and progress which was not up-held by many general practitioners. Instead, dissenting general practitioners viewed the 1990 contract as an instrument of control that, ironically if predictably, created instability by reducing practitioner autonomy and threatening the very "*nature of general practice*" (Stott 1994, Fugelli and Heath 1996).

Continuing and significant changes to who delivers primary care, and how it is delivered, makes defining the nature of general practice and its core values

increasingly difficult. Indeed, these very concepts have themselves become contentious (Heath 1995, Olsen 1996, Pringle and Heath 1997, RCGP Report 1996, Roland 1996, Kendrick and Hilton 1997). On the one hand, there is resistance to relinquish the ideal of general practitioners as autonomous providers of personal, holistic and generalist medical care, whilst on the other hand, there is a desire to develop roles as providers of increasingly specialised care, with the focus shifted from the individual to the practice population. While some GPs see the changing landscape as an opportunity for developing and extending specialist skills within general practice and forging links with other providers of primary and secondary care services (Kendrick and Hilton 1997), others have expressed concern that this will erode the traditional core skills of general practice (Heath 1995, Olsen and McMicheal 1998). Debates surrounding the role of general practitioners in implementing the new genetics can be interpreted in the context of these existing controversies. Genetic advances, and their proposed implementation by GPs, raises fundamental questions about the nature of general practice.

The new genetics has been described as "revolutionary" because it has the potential to transform not only the predictive, diagnostic and therapeutic dimensions of disease management, and existing models of risk communication, but also the underlying ideology fashioning health policy and health care provision (Rose, Lewontin and Kamin1990). The new genetics signifies a new level of reductionism for explaining disease aetiology. By focusing on individuals' genes as the prime cause of their ill health, their illness behaviour and experience, our collective attention is displaced from environmental, life-style , social and economic factors to focus on sequences of nucleotides that constitute each individuals' DNA. This has been described by a

prominent geneticist as the geneticisation of society, (Clarke 1995). At its extreme it may allow governments to evade responsibility for developing social policies aimed at creating healthy environments and promoting healthy behaviour. Biological determinism gives credibility to and extends the assumptions that underlie the belief that personal illness is determined through personal life style choices and behaviours which arise from within the individual alone. This in turn may affect the focus of scientific and medical research, which has implications for clinical practice as well as the future education and training of health professionals.

Biological Reductionism and the *new genetics*. The presentation of the new genetics in terms of a revolution involving the reformation of general practice by generating new tasks, detracts attention from its potential to preserve the core skills perceived to be under threat. The rhetoric of revolution inherent in the epithet new genetics obscures the extent to which the implementation of genetic research in general practice will need to draw on both traditional and new skills.

Rhetorics of revolution capture public imagination, and may also appeal to funding bodies, but must be regarded with suspicion. At what level is revolution implied? The new genetics, far from revolutionary, remain constant to the dominant force within western medicine for the past 400 years: reductionism. As a medical explanatory model, reductionism can be traced back to and extends, the Cartesian concept of body as machine, as part of which the body is conceived in terms of its constituent units. This concept is evident in our attempts to understand the body by analysing progressively smaller and smaller units; the history of medicine involves a series of ever greater anatomical reductions, from the dissection of the body to the

slicing of DNA. Can we assume that DNA will prove to be the final level of reduction or might yet more specific sites of biological process be identified? Admittedly, major advances in medicine have been firmly linked to the reductionist enterprise. Vaccines, blood groups and blood transfusions, immunology and transplant surgery, and the development of molecular pharmacotherapeutics are just a few successes.

However, reductionism has been linked to professionalisation and specialisation of knowledge, and thus communication problems between patients and doctors, as well as between medical specialists and generalists. Increasingly specialised knowledge within medicine creates intra-professional power hierarchies, and thus power (between the super-specialist, specialist and generalists) and widens the gap between health professionals and patients. To clarify this point I will provide a historical context to the power dynamics in the patient-doctor relationship and how challenges to power brought about change in some disciplines e.g. general practice. Several issues raised by genetic advances are discussed in this context e.g. biological determinism (genetic knowledge) with its implications for the classification and identification of both illness and the ill; certainty and risk; ethical dilemmas around confidentiality, disclosure, surveillance, screening, and the therapeutic gap. I will focus on the relationship between patient and general practitioner, an appropriate site because it draws upon my experience (personal and professional) and because the majority of the population in the U.K. are registered with a general practitioner who, in most instances, acts as the first point of contact for a patient. Other important features of general practice relevant to my discussions are its provision of continuous longitudinal care for individuals and families (Starfield 1994).

Why, when and with what expectations patients consult their doctor are shaped by medicine, culture and history, and so are usefully considered using anthropological frameworks. The nature of the patient-doctor relationship, its functions, boundaries and processes have been primarily determined by the medical profession. Controlling the patient-doctor relationship is part of a historical and continuing process by which doctors accumulate power/knowledge, authority, trust and autonomy for themselves and their profession. By the eighteenth century, medicine was defined by a network of practices, social structures and rituals. Knowledge about the methods, theories and practices of medicine could only be acquired by attending universities and joining the Royal Colleges—thus limiting medical knowledge to men from privileged classes (Sawday 1995). By the 19th century doctors had out-manoeuvred other contenders, e.g. apothecaries, birth-attendants and herbalists, to monopolise the right to practice medicine: manage illness, disease and child-birth (Daniel 1998). Doctors studied symptoms of the body, interpreting them using their knowledge of the body's chemical and physical anatomy. Symptoms were ordered to and classified in to larger groups—which according to Pierret (1995) created the subject of illness, making medicine a discourse on illness and not on the sick person. Jean Clavreul (1978) defined the patient's contribution as "the patient's role is simply to provide information on the state of the ailing body." The doctor's diagnosis, prognosis and management of an illness--the medical meaning given to a person's illness --was considered the most legitimate interpretation because medical reasoning was grounded in a scientific system considered to be objective, valid and reliable. Thus, by the twentieth century, power in the patient–doctor relationship lay with the doctor whose main focus was to normalise the abnormal body. It was not to understand the lived–experience of the sick person.

Genetic advances have implications for the balance of power between patient and doctor in the medical encounter, and have been presented as providing the medical profession with a new authority, certainty about the body and the diseases people will experience in their life time (Lupton 1998). Genetic advances have implications for the trust, discipline, and reputation of the medical profession, which according to Daniel (1998) maintain medical dominance and promote medical authority. The potential to know a patient's genome has been presented as providing doctors (and patients) with ultimate knowledge of the interior of the body—a process that started with dissection in the 16th century (Sawday 1995). The *new genetics* is the pinnacle of a process which began with the dissection of the body. The development of tests to identify genes predisposing people to cancer may function as another symbol, along with the white coat and the stethoscope, that provides validity and credibility to the doctor's diagnosis management and prognosis.

Daniel (1998) states that we put trust in practitioners because of our expectation of what that consultation and subsequent co-operation may achieve for us.
However, power may be eroded as patients recognise their GPs to be in-expert. Many people will not be familiar with genetic advances but are familiar enough to understand that it is a complex science and one which their GP may not be expert in (Emery, Kumar and Smith 1997). This has implications for the trust patients will have in the GPs' assessment of their genetic risks. If trust is in part a belief in the ability of an expert to understand and manage complexity and uncertainty then, as Daniel asserts, genetic advances may erode rather than build trust in the doctor, at least initially. Knowledge provided by genetic advances may be thought of as

revolutionary in that the knowledge/power relationship between patient and GP may be cancelled.

Only in the last third of the twentieth century did the process of contesting the sole right of the medical profession to intervene in illness and health begin. Works such as Illich's (1976) "Medical Nemsis" began to popularise the debate on the limits of technical medicine and its effectiveness and other works criticised the hold doctors had over the sick body and their lack of attention to the individual's lived-experience of illness and the medical encounter. Disciplines within medicine varied in their responses to such criticisms. In general practice, there was already considerable dissatisfaction with the bio-medical model's failure to manage the consequences of physical disease on people's social, emotional and psychological lives. Ignoring social and psychological issues were acknowledged to have a negative impact on patients' health outcomes (Ridsdale 1995). To address the lived-experience of illness GPs needed to engage with people's ideas, concerns and expectations of the illness and the medical encounter. The idea of holistic and humane clinical practice was aimed at addressing illness with reference to the patient's psychological and social context, and their experience of living with illness day-to-day. GPs attempts to achieve this are evident in the widely used definition of the general practitioner.

'*The general practitioner....his aim is to make early diagnoses. He will include and integrate physical, psychological, and social factors in his considerations about health and illness. He will make an initial decision about every problem which is presented to him as a doctor. He will undertake the continuing management of his patients with chronic, recurrent or terminal illnesses. He will know how and when to*

intervene through treatment prevention and education to promote the health of his patients and their families. He will recognise that he also has professional responsibility to the community.' (Leeuwenhorst Working Party 1974, Heath 1995 p.25)

The goal stated here of making *early diagnoses* fits well with the concept of genetic predisposition. However, the definition also implies a holistic approach which is at odds with the reductive and deterministic character of genetic medicine. The challenge then for practitioners is how to mediate between knowledge of a patient's potential genetic susceptibility to disease and their social, emotional and psychological contexts. The problem with such attempts at mediation is the scientific or medical uncertainty of how environment and genes interact to influence disease aetiology. As defined above, the general practitioner is a doctor who attempts to consider the contribution of nature (the gene) and nurture (social and environmental factors) to illness, the real with the constructed, the social with the biological. Paradoxically, genetic advances currently offer the general practitioner, not too much but, too little. This gap in knowledge compounds the problems raised later in the quoted definition about the GP's responsibility to the patient's family and community. For on the basis of current knowledge, how proactive should the GP be in advising patients to discuss their genetic risk with family members and partners?

Literature on the history of the patient-doctor relationship (Berne 1964, Byrne and Long 1976, Stott and Davis 1979, Helman 1994, Neighbour 1987) traces a shift away from a patriarchal, doctor-centred, task oriented relationship to more patient-centred and behaviour oriented relationships (Neighbour 1987). In the most

recent discussions the idea of a relationship is replaced by one of partnership, in which the doctor endeavours to understand the patient's agenda—which is often complex—and the patient's needs and preferences for information, and assistance with its interpretation and decision-making (Middleton and McKinley 2000). This suggests a process of democratisation about which we need to remain sceptical. It is doubtful whether, as Neighbour claims, "most doctors have now clambered down from their traditional pedestal." One reason why it is doubtful is because the style of consultation can leave underlying structures of power/knowledge uncontested. It is precisely these structures which are raised again by the genetic advances.

One area of change that has been noted in the patient-doctor relationship is the attempt to consider an individual's "social, psychological, cultural, intellectual and spiritual" contexts along side the physical (Royal College of GPs, Report 1996, vii). This attempt potentially conflicts with the geneticisation (Lippman 1991) of the relationship between patient and GP—a relationship which is reduced to one where the doctor is concerned primarily with what an individual's DNA can predict. For example, Aamra Darr (1997) notes that health professionals currently view consanguineous marriage negatively on the basis that off-spring of such marriages are at increased risk of homozygous recessive disorders such as thalassaemia. In this instance, genetics furnishes an up-to-date vocabulary for long-standing stigmaticisation. Quereshi (1997) notes the socially stabilising effect of consanguinity among British Pakistani and cautions genetic counsellors against blame and victimisation of parents. While genetic advances have tended to undermine the biological basis of 'ethnic' and particularly 'racial' difference, it also brings the danger of re-stigmatisation to those communities such as Ashkenazi Jewish women in

whom high rates of a specific mutation in the BRCA1:185delAG gene associated with breast cancer, are recognised but in whom Lerner (2001) reports there to be no increased incidence of the disease. It is possible to foresee scenarios where some general practitioners(GPs) may refuse to register groups of people who are associated high in put, high-cost therapies as was the case with people who were HIV positive.

The impact of genetic advances on the patient-doctor relationship will be determined by the understanding each party possess about the genes. Patients' understanding of genetics is shaped by popular culture e.g. life-style magazines, tabloids, soap operas, talk shows and sitcoms where genes are presented as explaining and determining diverse human traits e.g. obesity, criminality, homosexuality, shyness, intelligence, political beliefs and laziness. Such representations portray genes as powerful, deterministic and central to understanding ourselves (Nelkon and Lindee 1995). However, Nelkon and Lindee also found that some doctors believed strongly in genetic essentialism, a phenomenon that "reduces the self to a molecular entity, equating human beings in all their social, historical and moral complexity with their genes." We can speculate, that if both doctor and patient share deterministic views the relationship becomes fatalistic through such responses as 1) an over-estimation of risk—an emphasis on a gene as causing a specific disease e.g. breast cancer, is controversial as it gives the impression it acts independently of environmental forces which will influence its expression, 2) an underestimation of the individual's ability to influence their disease risk by changes in life style—fatalism (MacIntyre 1997), 3) radical prophylactic medical interventions, e.g. removal of healthy breasts and ovaries in women who have genetic susceptibility to these cancers. Such drastic interventions, are not always 100% successful due to incomplete removal of tissue,

and are contested in the medical literature on the grounds of their social and psychological costs to patients, many of whom might never have developed cancer (Lerner 2001). The fatalistic relationship can be seen as the culmination of the medical model of the patient-doctor relationship in which the illness --re-spelt as future illness-- and not the lived-experience of the patient, is the focus.

Genetic fatalism threatens to erode principles of patient-centredness, advocacy and confidentiality that belong to the partnership model of the patient-GP relationship (Heath 1995). It does so by reclassifying the currently healthy patient as the future-ill as well as subtly shifting the GP's perspective from individuals to their position in families; it raises problems of recording and disclosure of information that are not currently rationalised; it pressures confidentiality in the case of a GP who sees other members of the patient's family or who is approached by insurance agencies or other third parties. There is a increased potential for conflict and litigation between patients and GPs over disclosure.

Some GPs are reluctant to alert patients to their risk of genetic susceptibility to breast cancer because of the absence of effective genetic testing, screening technologies and therapies aimed a reducing risk of breast cancer or preventing the disease altogether. These GPs did not think bilateral mastectomy and oophrectomy were an appropriate management of risk because of the potential for immense physical, emotional and psychological harm to the patient (Kumar and Gantley 1999). The assumptions are problematic because they shift the relationship away from partnership (in which options and choices are discussed and decisions made together) to an older paternalistic model. Paradoxically, these doctors framed their reticence to discuss

genetic risk as 'ethical,' a use of that term which appears to refer to making decisions on the patient's behalf.

Lerner (2001) speaks of families in which young women who experience/or are aware that some of their close female relatives—mothers, sisters, aunts and grandmothers—have developed breast cancer. Many of these women will have knowledge of what the disease can do to their bodies and what doctors can do to their bodies in terms of radio- and chemotherapy as well as mastectomy. For these women the identification of BRCA 1 was at the same time empowering for instead of waiting for the disease to manifest they could find out their susceptibility through genetic testing. When a woman finds she carries the same genetic mutation as a relative with breast cancer then it is possible she may develop breast cancer at a similar age to her relative. What choices do such women have? At the moment there is no evidence that regular breast screening in younger women will detect early cancer, nor is there strong evidence that drugs such as tamoxifen will reduce the risk of her developing breast cancer. The one option surgeons have offered is "prophylactic mastectomy and oopherectomy"—the surgical removal of breasts and ovaries that are healthy. However, the apparent acceptability of this drastic procedure may be accounted for by a strong family history which suggests women's bodies are programmed to develop breast cancer and so they consider their breasts already diseased (Lerner 2001). However what is also known is that up 15 % of women who carry a BRCA1 or BRCA2 mutation associated with early onset breast cancer will not develop the disease. What the new genetics does not tell us is why and in whom? It could be action of beneficial factors in the environment or lifestyle of the patient or even other genes which are as yet unknown

to us, act to cancel out or modify effect of the breast cancer genes so they no longer cause breast cancer.

Currently, the new genetics lessens the power/knowledge structure because GPs who are not themselves informed or knowledgeable share with specialist an uncertainty over how such knowledge should be deployed and managed within specialist and generalist contexts. The new genetics is sufficiently novel that professional boundaries have not yet coalesced. Indeed, at present, the new genetics destabilises the premises of knowledge and power upon which the distinctions specialist, GP and patient are based. Perhaps only in this sense does it deserve the epithet revolutionary.

Rhetorics of responsibility Clarke (1995) has described the increasing reliance on genetic variation to explain individual differences as the geneticisation of society. At its extreme, this reliance may allow governments to evade responsibility for developing environmental, social, and economic policies necessary if one acknowledges the association of poverty, poor housing and unemployment with physical and psychological illness and disease. Rose, Lewontin, and Kamin (1990) have mapped the prioritisation of genetic research and knowledge over the past two decades with the implementation of health policies in which individual responsibility for health is presented as a moral, and at base economic, obligation. One might add here the promotion of private health care is an extension of this fundamentally economic rhetoric. Individual responsibility meant patients' commitment to securing health thorough the adoption of politically or medically sanctioned lifestyles and behaviour e.g. by stopping smoking, taking exercise, and decreasing saturated fat intake. There has been a gradual shift in responsibility for sustaining health from the

state to doctors and from doctors to individuals. Similarly genetic research focuses on the individual: indeed it is the culmination of a discourse of personal responsibility. Paradoxically, however, individual responsibility evaporates at the genetic level because, within the discourse of genetic determinism, an individual is seen to have no direct control over the fate of his or her health. The scientific validity of such a view is limited and fails to recognise the established links between environment, behaviour and phenotype (Piomin 1994)

Dilemmas from practice. Epidemiological evidence supports the role of behavioural, lifestyle, and environmental factors in assessing disease aetiology. Together with evidence of genetic contribution to disease, this indication renews the nature/nurture debate. The exact details of how gene expression may be modified by specific life styles and behavioural choices remains largely unclear in the context of common human diseases. While this gap in knowledge remains, GPs will be unable to provide tailored statistical estimates of risk incorporating lifestyle and genetic factors. This gap underlines the need to develop models of risk assessment that take in to account individual genotype, lifestyle and social and economic and environmental conditions. For GPs practising holistic medicine, it is precisely such links that will need to be considered. The new genetics intensifies the dilemma off how to combine knowledge produced within a deterministic framework with non-biological factors-social, psychological and cultural beliefs known to influence health and its management at many levels. For GPs, the potential danger of the new genetics lies in the prioritisation of biological information about the patient during clinical decision-making. For patients, the new genetics is potentially disempowering, and encourages fatalistic acceptance of behavioural, social, and environmental risks. Another paradox

is at work here. By over estimating an individual's predisposition to disease, the biologically deterministic model risks underestimating the individual's capacity to influence his or her medical history.

Policymakers and experts have spoken of the new genetics in terms of revolution and radical change for general practice. In adopting such rhetoric, they have either overlooked or not recognised the value of maintaining existing generalist knowledge and skills in implementing genetic advances. Their assumption that education, training, and provision of decision-support systems will produce a body of GPs willing to implement the new genetics cannot be taken for granted.

7.4 Combining qualitative and quantitative methods

In chapter three I examined the different philosophies of qualitative and quantitative research and methods, the challenges I faced in using both methodologies and how I negotiated these. I viewed the two methods as different but complementary and judged their usefulness in relation to their appropriateness for the questions being asked. Because qualitative and quantitative methods are fundamentally distinct (see chapter three) I did not think it was legitimate to create a hybrid method e.g. a closed questionnaire with a few open ended questions inviting GPs to write about their ideas and opinions. However, it was legitimate to "combine" both methods in the sense that each was used to answer different sets of questions concerning a single subject. In this work I wanted to understand and enumerate GPs' responses to the roles being advocated for them in relation to genetic advances and so it was appropriate to use both methods.

Thus qualitative and quantitative methods were not used to answer the same questions in some attempt at *triangulation* (Denzin 1970)—in other words I did not aim to use two different methods to try and replicate the findings of one by the other. The fact that the results of both studies concurred was not to be read as magnifying the validity of the findings or that the findings could be pooled to produce "one-truth." If there had been disagreement between the results of the two studies this would not have detracted from the validity of each study's findings. I concurred with the view that the results of one method can not validate nor falsify the findings of another method from a different paradigm because each must necessarily be addressing different sets of questions. For example, in this work each method was chosen because it was judged to be the best and most appropriate for addressing the questions being asked (see chapters four and six). The validity of each study was a reflection of the rigour inherent its design, data collection and analysis.

7.5 Summary of the strengths and weaknesses of the studies

The strength and weakness of the studies have been addressed in the body of the text-the aim here is to provide a brief summary. Overall, a major strength of this work comes from its cross disciplinary approach and its use of qualitative and quantitative methods to provide complementary knowledge of understanding GPs beliefs and decision-making and enumerating the frequency of specific practices and beliefs.

Specific strengths of the qualitative study relate to the careful exposition of data collection and analysis, respondent validation (chapter four), development of reflexivity, critical distance and theoretical sensitivity (chapters two and three), and an

attention to negative cases. Another strength of the study was the degree to which transferability was established (Lincoln and Guba 1985)—the extent to which the findings may be applied to similar contexts. When presenting the results to *average* GPs at local, national and international meetings I found GPs said our results fitted with their opinions and experiences in clinical practice. Another indication of transferability came from GPs' letters to the British Medical Journal which published the qualitative study. In these letters GPs said the findings echoed their opinions, ideas and experiences of genetics in clinical practice.

The qualitative study could be criticised for not developing a formal grounded theory. This was not possible within the resources allocated to the qualitative study. Another issue was that interviews with GPs were performed by another GP—this may be considered contentious by some because it raises the possibility that some assumptions and views held by general practitioners would be shared by the interviewer (see chapter four). Thus, it was possible that I may have overlooked or not explored with the necessary depth or critical distance some of the GPs' responses.

The quantitative study's main weaknesses was the small sample size which restricted data analysis to descriptive statistics and a limited factor analysis. Additionally, the sample was limited to the Wessex region which impaired the generalisability of the results. A strength of the GP survey was the relatively high response rate, and the fact that the questionnaire was developed from the qualitative data and so had a high degree of face and construct validity.

7.6 Section 3: Conclusions

The results of this work suggest that:

1) Many general practitioners do not believe that genetic testing for susceptibility to common conditions like breast cancer is likely to become a routine part of their clinical work in the near future.

2) Tensions exist between the role of general practitioners in implementing genetic advances identified by policy makers and that identified by general practitioners themselves.

3) The general practitioner is well placed to mediate between the biological and holistic models of health. General practitioners' ability to integrate patient experiences with genetic and other biological information is a key generalist skill given the potential of genetic advances to undermine the role of social, economic and environmental factors in disease aetiology.

4) The therapeutic gap was identified as a reason for not raising the issue of family history and genetic risk in the context of breast cancer.

5) Policy makers and experts can not assume that education and training will ensure GPs will implement genetic advances.

6) The hypothesis and findings of the qualitative study were upheld by the findings of the GP survey and the factor analysis of the data.

7.7 Future directions

This dissertation researched GPs' responses to identifying people with genetic susceptibility to common cancers. However, other members of the primary health care team e.g. practice nurses, health visitors and midwives, are likely to be involved. Their responses to identifying genetic susceptibility will require research and together with the GPs' responses will help clarify how genetic services and skills can be developed and organised in general practice.

My work examined individual GP's responses however, planning and organising the implementation of genetic advances requires research at other levels e.g. primary care trusts, which influence the services made available to populations based on perceptions of a population's needs and morbidity patterns. Thus, how a primary care trust prioritises developing or buying genetic risk assessment skills may differ from individual GPs, experts and policy-makers. Such differences will need to be understood.

GPs identified *genetic facilitators* as a way forward, in the short to medium term, for providing genetic risk assessment skills to groups of practices. This model has been successfully used in the context of single gene disorders and does merit research in the context of common conditions. Alongside research in to professionals' responses to organising and providing genetic services more research is required in to the impact genetic medicine will have on patients and their families.

These are just a few areas that will require investigation and which I hope I will be able to take forward in collaboration with others.

Appendix 1

Interview Guide

Dear Satinder, I decided that it might be useful to draw some of the issues from my observations of the GP course on genetics into a draft questionnaire. Please have a look at it and see if you think it makes sense, then we can discuss further. Then it might be worth thinking about undertaking an initial few interviews this month - Happy New Year! Madeleine [cc Ian]
5.1.95

Questionnaire for GPs on New Genetics

As you know, we have received funding from the RCGP to investigate the implications of research on 'new genetics' for general practice. We know that you recently took part in a course on medical genetics organised by the Wessex Regional Office of the RCGP and for this reason we would particularly like to hear your views. This is a little researched area, and our strategy is to combine qualitative and quantitative data. This interview is a part of the qualitative area of the study: the questions are deliberately general, and I would welcome any comments you feel are relevant.

This is not a test of your knowledge, but we intend to draw on your experience and expertise in order first to assess GP needs in relation to the new genetics, and then to offer practical help for GPs and their patients. At this stage we envisage the development of educational packages, including guidelines for the discussion of risk and specialist referral; however, we aim to use the information gained during these interviews to tailor both the nature of the package, and its contents, to GP needs. I'd like to start with a few background questions:

What do you understand by the "New Genetics"?

Who do you see as the key personnel in medical genetics?
[lead with the open question, encourage the respondent to reply as fully as possible; if necessary introduce the following as 'probes'; any new issues should be noted for inclusion in subsequent interviews]
- medical geneticists
- nurses
- the laboratory service

How do you see their roles?
How do you see their relationship with you as a GP?

What do you see as their specific expertise?
- the "tools" of genetics eg chromosomal, genetic, or DNA analysis, linkage studies, pedigree analysis
- offering expert counselling
- proactive screening

How well do you think you understand the potential and limitations of genetic susceptibility testing?

How do you see the contrasts between the service offered by medical geneticists, and that offered by a GP?
- short term meeting vs long term relationship with whole family

I'd like to move on now to ask some more specific questions.

Do you have any experience of referring patients to the medical genetics service. Could you identify the circumstances in which you would refer, and what information would you provide?
- during pregnancy, or planning a pregnancy
- after the birth of an affected[?] child
- if an older relative is diagnosed with an illness such as Huntington's chorea

Have any of your patients, as far as you know, had any contact with medical geneticists without being referred by you?
- for instance, some genetic nurses now try to make contact with families with specific conditions
- paediatrician referral

Have you found that patients have understood what they have been told? Some GPs have described part of their role as explaining what the geneticists have said: has this been your experience?

What sort of information have you received from the Medical Geneticists? Is there any other information that you would have found helpful?

The final section of questions relates to the future, and to some of the practical implications for GPs

Are you finding that patients are asking questions about the 'new genetics', or more generally about genetic risk?
- for instance, about family patterns of cancers

How do you respond to these questions? Do you feel that you have sufficient knowledge in this area? We know that you recently undertook a course in Medical Genetics: did this provide the kind of information you need?

What sort of records do you keep about the disease patterns within patients' families? Is this an area in which you would like help?
- pedigrees, the genogram
- is any information on causes of death in close family recorded, how?

Do you have concerns about ethical issues such as confidentiality of genetic information?
- around life insurance
- HIV
- providing information to 'high risk' patients, and to their families

Finally, I'd like to ask you to think more about the long term.

How do you foresee the future impact of the possiblity of individual genotyping on primary care?

- implications for lifestyle issues
- how to discuss risk factors (genetic and environmental)
- potential psychological effects of screening eg where prediction possible, but no treatment available; psychological impact of screening well people; impact on family relationships)
- confidentiality issues
- financial implications of 'purchasing' secondary care, or of accepting potentially expensive patients on to GP lists.
- developing the expertise of specific GPs, or Practice Nurses.

Thank you very much for your time. I have found our discussion very helpful, and we have covered all the issues on my agenda: is there anything else that you think, from your experience, that we should have included? ...

As I told you we hope to be developing some educational packages from this research: I would welcome your comments on these and would like, if I may, to send you a draft once this is available. Thank you...

All the bits in bold are comments from you, so need to be incorporated informally, as part of an informed discussion or 'guided conversation'. You need to make sure that the interview both covers the issues that you want to raise, and encourages informants to discuss and to add new comments, and flows as a conversation with a beginning, a middle and an end - You are in control (and informants expect you to have an agenda), but at the same time you are encouraging comment.

"GPs will have a pivotal role in providing genetic information"

There is a common assumption by policy makers that GPs will become increasingly involved in providing a primary care based genetic service. We are asking 200 GPs what they think. We would be grateful if you could spend 20 minutes on this short questionnaire. The answers will be confidential, and your annonymised results will be used to inform policy development. The findings of this study will be made available to you and presented at national forums.

1. Background Information

Please write down your:

a) Your sex (male or female) _____

b) The place and year you qualified _____

c) Your postgraduate qualifications _____

d) Your approximate list size _____

e) If you are a rural or urban practice _____

f) If you are a training practice, yes or no _____

Q . Please tick a box for each option. When a patient under the age of 50 years is diagnosed with cancer finding out about their family history of cancer....

a) Will give me ideas about the social problems that might arise in family

☐	☐	☐	☐	☐	☐
Strongly agree	agree	Neither agree nor disagree	disagree	strongly disagree	Don't know

b) Will **not** give me insight into the possible psychological problems that may arise

☐	☐	☐	☐	☐	☐
Strongly agree	agree	Neither agree nor disagree	disagree	strongly disagree	Don't know

c) Will provide explanation why the patient may have developed cancer

☐	☐	☐	☐	☐	☐
Strongly agree	agree	Neither agree nor disagree	disagree	strongly disagree	Don't know

d) Will give me genetic information about why the cancer developed

☐	☐	☐	☐	☐	☐
Strongly agree	agree	Neither agree nor disagree	disagree	strongly disagree	Don't know

e) Will give me information about the family's lifestyle risks that may explain the occurrence of cancer.

☐	☐	☐	☐	☐	☐
Strongly agree	agree	Neither agree nor disagree	disagree	strongly disagree	Don't know

f) Will allow me to assess if other family members are at genetic risk

☐	☐	☐	☐	☐	☐
Strongly agree	agree	Neither agree nor disagree	disagree	strongly disagree	Don't know

Appendix 5

Dear Dr

date

The impact of genetic advances on general practitioners

Advances made in medical genetics will require general practitioners to provide advice to patients on genetic susceptibility to common conditions such as cancer. Politicians and some clinical geneticists are very keen for GPs to take an active role in this. This questionnaire is a part of a bigger study which aims to explore GPs responses to the roles being identified for them. The questionnaire is about family history and cancer, and is short so should not take more than 15 minutes to complete.

The aim of this questionnaire is to collect information about you, your practice and how you might respond, in the context of collecting family history, when a patient under the age of 50 is diagnosed with cancer. Your responses will be anonymised. Taking part in this project will contribute to clarifying the role GPs can have in managing genetic risk for common conditions such as cancer.

If you have any queries about this questionnaire, or want more information then ring Dr. Satinder Kumar, at Aldermoor Health Centre on 01703-797731, or e-mail at skk@soton.ac.uk. Thank you very much for your time.

Yours sincerely

Dr Satinder Kumar
(General Practitioner).

Time Table of Key Events

Appendix 4

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